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## **Medical consequences of pathogenic CNVs in adults: Analysis of the UK Biobank**

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## ABSTRACT

**Background** Genomic copy number variants (CNVs) increase risk for early-onset neurodevelopmental disorders but their impact on medical outcomes in later life is still poorly understood. The UK Biobank allows us to study the medical consequences of CNVs in middle and old age in half a million well-phenotyped adults.

**Methods** We analysed all Biobank participants for the presence of 54 CNVs associated with genomic disorders or clinical phenotypes, including their reciprocal deletions or duplications. After array quality control and exclusion of first-degree relatives, we compared 381,452 participants of white British or Irish origin who carried no CNVs, with carriers of each of the 54 CNVs (ranging from five to 2843 persons). We used logistic regression analysis to estimate the risk of developing 58 common medical phenotypes (3132 comparisons).

**Results and Conclusions** Many of the CNVs have profound effects on medical health and mortality, even in people who have largely escaped early neurodevelopmental outcomes. Forty-six CNV-phenotype associations were significant at a false discovery rate (FDR) threshold of 0.1, all in the direction of increased risk. Known medical consequences of CNVs were confirmed, but most identified associations are novel. Deletions at 16p11.2 and 16p12.1 had the largest numbers of significantly associated phenotypes (seven each). Diabetes, hypertension, obesity and renal failure were affected by the highest numbers of CNVs. Our work should inform clinicians in planning and managing the medical care of CNV carriers.

## INTRODUCTION

Genomic copy number variations (CNVs) are structural alterations to chromosomes of >1,000 bases in length that can intersect multiple genes.[1] Specific CNVs have been shown to increase risk for autism spectrum disorders,[2] developmental delay and other neurodevelopmental disorders,[3] and schizophrenia.[4] Apart from their association with neurodevelopmental and psychiatric outcomes, these CNVs can lead to medical disorders. Several CNVs, for example deletions at 22q11.2,[5] have been extensively studied on hundreds of carriers and their medical consequences are well established. However, for CNVs with lower penetrance, very rare CNVs, or several reciprocal deletions/duplications of known genomic disorders, the associated medical phenotypes have not been identified. Moreover, most research has been performed on children and young people referred to genetic clinics,[3, 6] creating a strong referral bias towards recording high rates of developmental delay, early-onset medical conditions and more adverse outcomes. Most CNVs display incomplete penetrance,[7] resulting in apparently unaffected adult carriers in the general population. The rate of medical outcomes in later life of CNV carriers, or in the general population as a whole, has not been addressed in adequately powered studies to date.

The establishment of the UK Biobank presents a unique opportunity to examine the spectrum of medical outcomes of CNVs in middle- and old-aged people, as all half a million participants have been assessed with identical methods and blindly to their CNV status. The Biobank collects longitudinal data from hospital admissions, self-report, death certificates, cancer registries and primary care (general practitioners') records. Here, we report on the medical consequences of carrier status for 54 CNVs that are recognised as associated with clinical phenotypes or genomic disorders,[3, 6, 8] including their reciprocal deletions/duplications.

## METHODS

**Participants.** The UK Biobank recruited just over half a million people from the general population of the UK, using National Health Service (NHS) patient registers, with no exclusion criteria. Participants have consented to provide personal and health information, urine, saliva and blood samples, and to have their DNA tested. We obtained approval from the UK Biobank to analyse the CNVs in project 14421: “Identifying the spectrum of biomedical traits in adults with pathogenic copy number variants (CNVs)”.

Participants were between 40 and 69 years of age at the time of recruitment between 2006 and 2010. As the lifetime prevalence of disorders often varies by ancestry, we restricted the analysis to those participants who declared themselves as “white British or Irish”: 421,268 participants who passed our genotyping QC filters (see below). After exclusion of first-degree relatives, 396,725 subjects were retained for analysis, 53.8% of whom were female. The mean age at the end of the current follow-up interval for medical outcomes (in 2016) was 64.7 years, SD = 8.0 years.

**CNV calling.** Samples were genotyped at the Affymetrix Research Services Laboratory, Santa Clara, CA on two arrays with 95% common content between them: around 50,000 samples were genotyped on the UK BiLEVE Array (807,411 probes), and the remaining samples on the UK Biobank Axiom Array (820,967 probes).[9] We downloaded the anonymised genotypic data from the UK Biobank as 488,415 raw (CEL) files and analysed them with the methods we reported previously.[10] Briefly, we generated normalised signal intensity data, genotype calls and confidences, using ~750,000 biallelic markers. These were then processed with PennCNV-Affy software.[11] Individual samples were excluded if they had >30 CNVs, a waviness factor >0.03 or <-0.03, a call rate <96%, or Log R Ratio standard deviation >0.35. A total of 25,069 files were excluded after this QC (5.1%). Individual CNVs

were excluded if they were covered by <10 probes or had a density coverage of less than one probe per 20,000 base pairs.

**Choice of CNVs.** We compiled a list of 92 CNVs in 47 genomic locations from two widely accepted sources that proposed largely overlapping sets of CNVs (Supplementary Table 1 in Supplementary material).[3, 6] The authors of these studies used information from databases, reviews and publications to produce lists of CNV regions that lead to genomic disorders, congenital malformations, neurodevelopmental or other clinical phenotypes. We refer to this set of 92 CNVs as “pathogenic”, consistent with the criteria proposed by the American College of Medical Genetics standards which describe as pathogenic those CNVs that have been documented as clinically significant in multiple peer-reviewed publications, even if penetrance and expressivity of the CNV are known to be variable.[12] Many (but not all) have been shown to statistically increase risk for developmental delay.[3] Supplementary Table 1 lists the sources for selection and our criteria for inclusion in analysis. Several overlapping or adjacent CNVs listed as separate loci in the original publications were grouped together (e.g. the “small” and the “common” 22q11.2, or the “small” and the “large” 16p13.11 deletions/duplications). As a rule, the reciprocal deletions/duplications of known genomic disorders were also included by the above authors and by us, in order to examine their medical consequences, even if the evidence for their pathogenicity has not been established.

The criteria for calling CNVs that do not span the full critical region are given in Supplementary Table 2. As a rule, a CNV had to intersect at least 50% of the critical region, marked as “Location (hg19)”, and intersect the relevant candidate genes, if known. For single gene CNVs, we required deletions to intersect at least one exon, and duplications to span the whole coding region, as the functional consequences of partial gene duplications can be

unpredictable, while deletions of any part of the coding sequence of a gene are likely to act as loss-of-function mutations. We observed several loci, mostly telomeric, where a number of small CNVs were preferentially called on arrays that failed QC (marked “Unreliable” in Supplementary Table 1). We excluded these loci from analysis in order to avoid potential false-positives on this genotyping platform. We also excluded from analysis CNVs with fewer than five observations in the full sample, as being too rare for statistical analysis (marked “Rare” in Supplementary Table 1). The above filtering left 54 CNVs for analysis (Table 1).

**Table 1. List of 54 CNVs analysed in this study.** The column “N sign. FDR=0.1” shows the number of significant associations between the CNV and medical phenotypes at a threshold of FDR = 0.1. Further details in Supplementary Table 1. First-degree relatives are included in the numbers of carriers.

CNV locus	Location (hg19)	N genes	N carriers (%)	N sign. FDR=0.1
TAR del	chr1:145,39-145,81	17	75 (0.018)	0
TAR dup	chr1:145,39-145,81	17	436 (0.1)	1
1q21.1del	chr1:146,53-147,39	9	113 (0.027)	2
1q21.1dup	chr1:146,53-147,39	9	177 (0.042)	1
<i>NRXN1</i> del	chr2:50,14-51,26	1	163 (0.039)	1
2q11.2del	chr2:96,74-97,68	22	31 (0.007)	0
2q11.2dup	chr2:96,74-97,68	22	29 (0.007)	0
2q13del( <i>NPH1</i> )	chr2:110,86-110,98	3	2448 (0.58)	0
2q13dup( <i>NPH1</i> )	chr2:110,86-110,98	3	1976 (0.47)	0
2q13del	chr2:111,39-112,01	3	53 (0.013)	0
2q13dup	chr2:111,39-112,01	3	71 (0.017)	1
2q21.1del	chr2:131,48-131,93	5	41 (0.01)	0
2q21.1dup	chr2:131,48-131,93	5	59 (0.014)	0
3q29del	chr3:195,72-197,35	28	9 (0.002)	0
3q29dup	chr3:195,72-197,35	28	5 (0.001)	6
WBS dup	chr7:72,74-74,14	26	14 (0.003)	0
7q11.23dup_distal	chr7:75,14-76,06	16	24 (0.006)	0
8p23.1dup	chr8:8,10-11,87	35	6 (0.001)	0
10q11.21q11.23del	chr10:49,39-51,06	19	57 (0.014)	0
10q11.21q11.23dup	chr10:49,39-51,06	19	43 (0.01)	0
10q23dup	chr10:82,05-88,93	29	7 (0.002)	0
13q12del( <i>CRYL1</i> )	chr13:20,98-21,10	2	379 (0.09)	0
13q12dup( <i>CRYL1</i> )	chr13:20,98-21,10	2	10 (0.002)	0

13q12.12del	chr13:23,56-24,88	10	85 (0.02)	0
13q12.12dup	chr13:23,56-24,88	10	236 (0.056)	0
15q11.2del	chr15:22,81-23,09	5	1664 (0.39)	0
15q11.2dup	chr15:22,81-23,09	5	2041 (0.48)	0
PWS_dup	chr15:23,68-28,39	116	19 (0.005)	0
15q11q13del_BP3-BP4( <i>APBA2, TJP</i> )	chr15:29,16-30,38	4	16 (0.004)	1
15q11q13dup_BP3-BP4( <i>APBA2, TJP</i> )	chr15:29,16-30,38	4	53 (0.013)	0
15q11q13dup_BP3-BP5	chr15:29,16-32,46	17	9 (0.002)	0
15q13.3del	chr15:31,08-32,46	8	42 (0.01)	2
15q13.3dup	chr15:31,08-32,46	8	240 (0.057)	0
15q13.3del( <i>CHRNA7</i> )	chr15:32,02-32,46	1	10 (0.002)	0
15q13.3dup( <i>CHRNA7</i> )	chr15:32,02-32,46	1	3031 (0.72)	0
15q24dup	chr15:72,90-78,15	77	9 (0.002)	0
16p13.11del	chr16:15,51-16,29	7	131 (0.031)	1
16p13.11dup	chr16:15,51-16,29	7	828 (0.2)	2
16p12.1del	chr16:21,95-22,43	8	246 (0.058)	7
16p12.1dup	chr16:21,95-22,43	8	202 (0.048)	0
16p11.2distal_del	chr16:28,82-29,05	11	58 (0.014)	3
16p11.2distal_dup	chr16:28,82-29,05	11	137 (0.033)	0
16p11.2del	chr16:29,65-30,20	30	110 (0.026)	7
16p11.2dup	chr16:29,65-30,20	30	138 (0.033)	2
17p12del(HNPP)	chr17:14,14-15,43	8	237 (0.056)	1
17p12dup(CMT1A)	chr17:14,14-15,43	8	124 (0.029)	3
Potocki-Lupski Syndrome	chr17:16,81-20,21	59	5 (0.001)	0
17q11.2del( <i>NFI</i> )	chr17:29,12-30,27	19	9 (0.002)	0
17q12del	chr17:34,81-36,22	17	9 (0.002)	2
17q12dup	chr17:34,81-36,22	17	101 (0.024)	1
22q11.2del	chr22:19,04-21,47	61	10 (0.0024)	0
22q11.2dup	chr22:19,04-21,47	61	280 (0.066)	2
22q11.2distal_del	chr22:21,92-23,65	26	5 (0.001)	1
22q11.2distal_dup	chr22:21,92-23,65	26	13 (0.003)	0

**Choice of medical phenotypes.** Data on health outcomes were collected from several sources. Self-declared illnesses were disclosed by participants at their initial assessments and coded into 445 distinct categories. Hospital discharge diagnoses (primary and secondary) and death certificates contain over 11,000 ICD10 codes assigned to at least one participant. Analysing each individual code separately against 54 CNV loci would result in small numbers of participants with each code and fail to provide the statistical power needed to detect true associations. To reduce the dimensionality of the data and therefore increase power and provide more meaningful results, we grouped together discrete disease entities into broader disease groups. A participant was coded as a “case” if he/she had a relevant diagnosis on at least one occasion, in any of the above sources of information. We gave preference to common conditions and grouped disorders into recognised categories, based on



organ, system, or aetiology, while excluding from the current analysis infectious diseases, injuries, and neuropsychiatric disorders (the latter being analysed separately). The disease codes used to construct each phenotype group are listed in Supplementary Table 3. For myocardial infarction and stroke, we used the “adjudicated” data provided by the UK Biobank (data fields 42000 to 42013). Phenotype groups found in fewer than 2000 participants were not included. The final list of disease groups contains 58 entities, including “death during follow-up” obtained from the death registries. Data on cancer were taken only from the UK cancer registries, as collected and supplied by the UK Biobank, as this is the most reliable and complete resource for cancers in the UK. For the current work we considered all malignant cancers as a single phenotype. As risk for cancer was not significantly affected by CNVs as a group, and because most individual cancers affected relatively small numbers of patients, we did not analyse the cancers further by sub-type.

**Statistical analysis.** Analyses were performed in the statistical package R (version 3.3.2) using a Linux server. We examined the effect of the presence of a CNV on each medical phenotype with logistic regression analysis. As co-variates we used age, gender, array type (Axiom/BiLEVE), Townsend deprivation index (as a measure of the socioeconomic status) and the first 15 principal components from the genetic analysis, as provided by the UK Biobank. We used Firth's bias-reduced logistic regression method,[13] with the R library “logistf”, as it better handles cells with small numbers. We report the resulting p-values, odds ratios (ORs) and 95% confidence intervals for the ORs. We also report the uncorrected Relative Risk (RR), for having the phenotype in carriers of a specific CNV and non-carriers of any of the 54 CNVs. (RR is used for the additional images on our website (<http://kirov.psychm.cf.ac.uk/>), as it returns the more intuitive value of zero for associations with zero CNVs in cases.) Conservative Bonferroni correction for the testing of 54 CNVs × 58 phenotypes, gives a  $p < 1.6 \times 10^{-5}$  as a project-wide significance level. As many true-positive

associations were expected, it is more appropriate to use the Benjamini-Hochberg false discovery rate (FDR) for correction of p-values.[14] Our preferred false discovery rate (B-H FDR) is 0.1.

## **RESULTS AND DISCUSSION**

**Quality control.** The Affymetrix arrays produced reliable calls for the 54 CNVs. This is not surprising, given the large size and good probe coverage of these CNVs. This impression is confirmed by the remarkably similar CNVs frequencies, compared to those reported by us in previous control populations (Supplementary Table 4 and Supplementary Figure 1). There were no apparent batch effects affecting the calls: the distribution of each CNV in the 106 batches produced no outliers from the expected Poisson distribution, after taking into account the multiple testing for 54 CNVs (Supplementary Table 5). The best confirmation of the data quality would be the identification of well-known phenotypes associated with specific CNVs. This was indeed the case (Table 2), as we identified, for example, the known associations of neuropathies and 17p12 deletions / duplications;[15] obesity and deletions at 16p11.2 and 16p11.2 distal;[16, 17] diabetes and 17q12 deletions (also called “renal cysts and diabetes syndrome”).[18] This increases our confidence that the newly identified associations are also real.

**Effects of CNVs on medical phenotypes.** Each of the 54 CNVs was tested for association with each of the 58 medical phenotypes (a total of 3132 tests). Results are presented as odds ratios (ORs) for risk of developing the phenotype, corrected for age, sex and the other co-variables detailed in the Methods. All results are presented in Supplementary Table 6 (grouped by CNV) and in Supplementary Table 7 (grouped by phenotype).

The top 14 significant phenotype/CNV associations (Table 2) survive a Bonferroni correction for 3132 tests (a project-wide significant p-value threshold of  $1.6 \times 10^{-5}$ ). This correction is over-conservative, due to medical co-morbidities (e.g. people with diabetes also have increased rates of heart attacks, stroke and others). A more appropriate correction of statistical significance for this analysis is the Benjamini-Hochberg FDR.[14] There are 46 CNV/phenotype comparisons that were significant at a FDR = 0.1 (Table 2). Most of these are novel associations and none are protective for the tested phenotypes (all have OR > 1).

**Table 2. CNV/Phenotype associations significant at FDR=0.1.** The numbers of cases and controls are the numbers of people who have the phenotype, while the number of CNV carriers is presented in brackets. The “Expected number of CNVs in cases” is extrapolated from their frequencies in the controls. Uncorrected p-values and Benjamini-Hochberg FDR corrected p-values for 3132 tests (“p-value B-H FDR”) are also shown. OR and 95% confidence interval of the OR are produced by Firth’s logistic regression analysis (Methods). More details in Supplementary Tables 6 and 7. “Known finding” refers to known medical consequences, listed in Supplementary Table 1, or to phenotypes that appear a consequence of the known ones (marked as “secondary” in the table, as discussed below)

CNV	Phenotype	N con (N CNVs)	N case (N CNV)	Expected N CNVs in cases	P-value	P-value B-H FDR	OR (95%CI)	Known finding
TAR dup	obesity	372000 (385)	9860 (23)	10.2	0.00054	0.047	2.3 (1.5-3.4)	
1q21.1 del	heart failure	376477 (100)	5081 (6)	1.3	0.0011	0.083	5.3 (2.1-11.2)	Yes
1q21.1 del	cataract	359694 (92)	21864 (14)	5.6	0.00047	0.042	3.2 (1.7-5.6)	Yes
1q21.1 dup	diabetes, type 2	360864 (146)	20756 (22)	8.4	0.00017	0.025	2.7 (1.6-4.1)	
NRXN1 del	aneurism	379638 (152)	1971 (5)	0.8	0.00042	0.041	7.6 (2.8-16.4)	
2q13 dup	diabetes, type 2	360778 (60)	20745 (11)	3.5	0.0012	0.094	3.4 (1.7-6.3)	
3q29 dup	any cancer	331757 (1)	49700 (4)	0.15	$6.23 \times 10^{-5}$	0.011	37.5 (6.5-389.1)	
3q29 dup	diverticular disease intestine	354321 (2)	27136 (3)	0.2	0.0001	0.017	41.8 (7.4-276.0)	
3q29 dup	inflammatory bowel disease	362864 (2)	18593 (3)	0.1	0.00013	0.021	35.5 (6.7-217.8)	
3q29 dup	renal failure	373535 (3)	7922 (2)	0.1	0.00022	0.027	58.4 (9.2-324.8)	
3q29 dup	death	370486 (3)	10971 (2)	0.1	0.0013	0.093	27.8 (4.5-146.0)	
15q11q13 del BP3-BP4	gastric reflux	347165 (8)	34301 (6)	0.8	0.00018	0.025	9.1 (3.1-25.4)	
15q13.3 del	diabetes, type 2	360746 (28)	20743 (9)	1.6	0.00038	0.039	4.9 (2.2-10.2)	
15q13.3 del	asthma	332151 (23)	49338 (14)	3.4	0.00018	0.026	3.9 (2.0-7.4)	
16p13.11 del	obesity	371729 (114)	9847 (10)	3.0	0.0013	0.096	3.4 (1.7-6.2)	
16p13.11 dup	hypertension	261304 (483)	120931 (300)	223.5	$2.05 \times 10^{-5}$	0.0043	1.4 (1.2-1.6)	

16p13.11 dup	death	371226 (743)	11009 (40)	22.0	0.00097	0.080	1.8 (1.3-2.4)	
16p12.1 del	obesity	371827 (212)	9860 (23)	5.6	$1.11 \times 10^{-7}$	$4.95 \times 10^{-5}$	4.0 (2.5-6.0)	
16p12.1 del	hypertension	260945 (124)	120742 (111)	57.4	$8.64 \times 10^{-8}$	$5.41 \times 10^{-5}$	2.1 (1.6-2.8)	
16p12.1 del	renal failure	373750 (218)	7937 (17)	4.6	$7.23 \times 10^{-6}$	0.0021	3.8 (2.3-6.1)	
16p12.1 del	diabetes, type 2	360926 (208)	20761 (27)	12.0	0.00021	0.028	2.3 (1.5-3.5)	
16p12.1 del	heart other	369848 (217)	11839 (18)	6.9	0.00029	0.034	2.8 (1.7-4.4)	Yes
16p12.1 del	ureter / bladder	333111 (186)	48576 (49)	27.1	0.00033	0.036	1.9 (1.3-2.5)	
16p12.1 del	respiratory	360911 (209)	20776 (26)	12.0	0.00069	0.059	2.2 (1.4-3.2)	
16p11.2 distal del	gout	374410 (48)	7096 (6)	0.9	0.00046	0.042	6.5 (2.5-14.4)	
16p11.2 distal del	obesity	371660 (45)	9846 (9)	1.2	$1.14 \times 10^{-5}$	0.0028	7.1 (3.3-13.7)	Yes
16p11.2 distal del	diabetes, type 2	360757 (39)	20749 (15)	2.2	$8.86 \times 10^{-8}$	$4.63 \times 10^{-5}$	7.0 (3.7-12.6)	
16p11.2 del	diabetes, type 2	360794 (76)	20761 (27)	4.4	$2.54 \times 10^{-11}$	$3.98 \times 10^{-8}$	6.1 (3.8-9.5)	Secondary
16p11.2 del	obesity	371699 (84)	9856 (19)	2.2	$7.39 \times 10^{-10}$	$7.71 \times 10^{-7}$	6.8 (4.0-11.0)	Yes
16p11.2 del	anaemia	362396 (84)	19159 (19)	4.4	$2.15 \times 10^{-6}$	0.00075	4.0 (2.4-6.5)	
16p11.2 del	asthma	332199 (71)	49356 (32)	10.5	$1.33 \times 10^{-5}$	0.0030	2.7 (1.8-4.1)	
16p11.2 del	renal failure	373625 (93)	7930 (10)	2.0	$6.04 \times 10^{-5}$	0.012	5.1 (2.5-9.5)	
16p11.2 del	hypertension	260873 (52)	120682 (51)	24.1	$9.44 \times 10^{-6}$	0.0025	2.6 (1.7-3.8)	Secondary
16p11.2 del	osteoarthritis	312820 (73)	68735 (30)	16.0	0.00031	0.035	2.4 (1.5-3.6)	Secondary
16p11.2 dup	irritable bowel syndrome	368567 (118)	13016 (13)	4.2	0.00036	0.037	3.3 (1.8-5.7)	
16p11.2 dup	sciatica	338444 (103)	43139 (28)	13.1	0.001	0.083	2.1 (1.4-3.2)	
17p12 HNPP del	neuropathies	365743 (194)	15928 (25)	8.4	$5.44 \times 10^{-6}$	0.0017	3.0 (2.0-4.5)	Yes
17p12 CMT1A dup	neuropathies	365609 (60)	15959 (56)	2.6	$3.9 \times 10^{-124}$	$1.2 \times 10^{-120}$	21.8 (15.0-31.5)	Yes
17p12 CMT1A dup	anaemia	362411 (99)	19157 (17)	5.2	$6.7 \times 10^{-5}$	0.012	3.3 (1.9-5.4)	
17p12 CMT1A dup	stroke	372741 (107)	8827 (9)	2.5	0.0013	0.094	3.7 (1.8-6.9)	
17q12 del	diabetes insulin dependent	378861 (3)	2598 (4)	0.0	$3.93 \times 10^{-8}$	$3.08 \times 10^{-5}$	135.9 (31.2-641.1)	Yes
17q12 del	digestive	299609 (1)	81850 (6)	0.3	0.00044	0.042	15.4 (3.2-150.3)	
17q12 dup	renal failure	373622 (90)	7929 (9)	1.9	0.00029	0.035	4.6 (2.2-8.6)	
22q11.2 dup	hernia	333779 (215)	47939 (51)	30.9	0.0013	0.094	1.7 (1.2-2.3)	
22q11.2 dup	gastric reflux	347374 (217)	34344 (49)	21.5	$1.82 \times 10^{-6}$	0.0007	2.3 (1.7-3.1)	
22q11.2 distal del	aneurism	379490 (4)	1967 (1)	0.0	0.0013	0.092	104.9 (9.7-673.6)	

A total of 330 tests were nominally significant (at  $p < 0.05$ ), instead of the expected 157.

Figure 1 shows the distribution of p-values, with a clear trend for over-representation below the  $p < 0.1$  level. This suggests that there are many more real associations, than those presented in Table 2, but they cannot be identified with sufficient statistical significance in a sample of this size. Clinicians might therefore decide to also consider consequences of CNVs that do not survive our corrections.

Figure 1 about here

Deletions at 16p11.2 and 16p12.1 had the largest numbers of significantly associated phenotypes (seven each). Deletions at 16p11.2 are a known risk factor for obesity.[16] We now provide data showing that adult carriers also have a high incidence of diabetes, osteoarthritis and hypertension, possibly as expected consequences / comorbidities of obesity. Other associated phenotypes are not necessarily linked to a high BMI, such as asthma, anaemia and renal problems, suggesting that this and other CNVs have pleiotropic effects (see conditional analysis below). This should be expected from CNVs intersecting multiple genes. This has already been shown for some large CNVs, e.g. 22q11.2 deletions, where highly variable phenotypic presentations are the norm.[5]

We should point out that CNVs with higher numbers of significant results are not necessarily the most pathogenic ones, as significance depends also on CNV frequency, which is low for the most pathogenic CNVs in this population. Such CNVs are under-represented in the UK Biobank, as the participants are middle-aged and participation is subject to “healthy volunteer” selection bias.[19] For example 22q11.2 deletions are highly pathogenic,[5] but there were only 10 such carriers in the Biobank, instead of the expected ~100 (the rate of this deletion among newborns is ~1:4,000).[7] These 10 carriers were not sufficient to produce significant results at  $FDR = 0.1$ , even for  $ORs > 10$  (Supplementary Table 6). The more

informative data from our research is on CNVs with lower penetrance, as they are more common.

The increased risk for medical morbidities or mortality observed in CNV carriers is unlikely to be due to the presence of early neurodevelopmental disorders or schizophrenia in carriers, as the UK Biobank population has largely escaped such conditions: only 34 of the 14,791 people who had one of the tested CNVs had schizophrenia, 17 had developmental delay and four had autism. Accidental death or death in epilepsy can not account for the increased death rate in CNV carriers: out of the 504 CNV carriers who had died during follow-up, only one had “Sudden unexpected death in epilepsy” and another four had accidental deaths (motor/pedal cyclist accidents and falls from a high place). All death causes in CNV carriers, according to the death registries, are listed in Supplementary Table 8.

**Phenotypes most likely to be affected by CNVs.** Diabetes, hypertension, obesity and renal failure were the phenotypes affected by the highest number of CNVs (Table 2). The real number of affected phenotypes by the CNVs is probably much higher, as suggested in Figure 1. We can provide further evidence for this, by testing the effect on the phenotypes in the group of pathogenic CNV carriers as a whole, thus substantially increasing the statistical power. After excluding the five common CNVs (as they would determine the results due to their high frequencies), the remaining 4782 carriers of 49 rare CNVs, had significantly increased risk for developing 26 of the 58 tested phenotypes (Figure 2). Hypertension, diabetes, cardiac, respiratory and renal disorders dominate the top results. These are common phenotypes that increase mortality. We do indeed observe an increased death rate among CNV carriers during the follow-up period of Biobank participants (death was the second most-significant phenotype, Figure 2). The relative risk (RR) of death from each CNV is presented in Figure 3, where the RRs are ordered by the statistical strength of the association

(strongest p-value on the left). The vertical line demarcates the 12 CNVs that are nominally significantly associated with increased mortality ( $p < 0.05$ ). Not surprisingly, the more pathogenic CNVs were also associated with increased mortality. The top significant CNV was, unexpectedly, the relatively common duplication at 16p13.11, found in ~0.2% of the general population, an association that has not been outlined before.

Figures 2 and 3 about here

Most of the reported associations are novel, although some of them can be explained as logical adult medical consequences of known, early-onset phenotypes, e.g. obesity leading to diabetes, hypertension and increased cardiovascular mortality. In order to test this possibility, we performed a conditional analysis of three CNVs and two phenotypes, where obesity is most likely to account for some or all of the associations, by adding the body mass index (BMI) as a new co-variate to the original analysis. This analysis amounted to 276 independent tests, to which we applied again the Benjamini-Hochberg FDR method, to establish which associations remained significant at  $FDR = 0.1$ , after controlling for BMI. Obesity is a well-established phenotype of 16p11.2 classic and distal deletions. The results and comparisons with the original analysis for all phenotypes and these two CNVs are shown in Supplementary Tables 9 and 10 and Supplementary Figures 2 and 3. For 16p11.2 classic deletion, four of the six originally significant associations at  $FDR = 0.1$  remained significant (excluding obesity from these numbers). The changes in the ORs give a better global impression of the changes (Supplementary Figure 2), and indicate that several associations are much reduced: diabetes type 1 and 2, hypertension, high cholesterol, gout and osteoarthritis. This indicates that these disorders are, to a large extent, consequences of obesity. However, the ORs for anaemia and asthma didn't change substantially. 16p11.2 distal deletions showed smaller reductions in the ORs (Supplementary Figure 3) and four



phenotypes (excluding obesity) remain significant at FDR=0.1. This pattern suggests that other factors also play a role in the causation of phenotypes in carriers of this CNV. Although deletions at 16p12.1 have not been an established cause for obesity, the pattern of results (Table 2) also raised the question as to whether the multiple associated phenotypes could be explained by obesity. Therefore we included this CNV in the conditional analysis (Supplementary Table 11 and Supplementary Figure 4). Increased BMI appeared to play a smaller role in the causation of disease phenotypes for this CNV, with small changes in the ORs and the number of significant results.

Somewhat counterintuitively, the association with obesity does not get fully abolished when the analysis is corrected for BMI. There are, however several factors that can explain this apparent anomaly. Most relevantly, the phenotype “Obesity” is not equivalent to high BMI. It is a hospital ICD10 diagnosis, made on a small proportion of people who have a BMI>30. In fact, 24.3% of the Biobank population has a BMI>30, qualifying them for a diagnosis of Obesity, but only 9.2% of them received this diagnosis. Furthermore, Obesity is a categorical variable, while BMI is a continuous one, making them not equivalent from a statistical point of view, and therefore adjusting an analysis of one for another does not necessarily remove all evidence for association. The distribution of BMI values is very different in the three CNVs tested: 71.6% of 16p11.2 deletion carriers had a BMI>30, compared to 55.6% of 16p11.2 distal deletion carriers and 37% of 16p12.1 deletion carriers (Supplementary Figure 5 a-c). ICD10 diagnosis of “Obesity” was given to correspondingly smaller proportions of carriers: 18.6%, 16.7% and 9.8%. These differences could explain why correcting for BMI does not lead to identical changes to the associations of the three CNVs.

We also tested whether increased BMI accounted for associations of diabetes type 2 or mortality with any of the 54 CNVs (Supplementary Tables 12 and 13 and Supplementary

Figures 6 and 7). As already reported above, this was the case for diabetes and the “classic” and “distal” 16p11.2 deletions. However, for 1q21.1 and 2q13 duplications, 22q11.2 distal deletions and 17q12 deletions (also known as “renal cysts and diabetes syndrome”) the ORs for diabetes increased, suggesting that these CNVs have a more direct effect on the development of diabetes. In total, six CNVs were significantly associated with diabetes, after controlling for BMI (Supplementary Table 12). The associations with mortality remained essentially unchanged after correction with BMI, with four significantly associated CNVs (Supplementary Table 13) and very similar ORs (Supplementary Figure 7), indicating that obesity is only one of many consequences that shortens the lives of CNV carriers.

**Homozygous deletions and more than one CNV per person.** Only four carriers of homozygous deletions were found, perhaps not surprisingly for this relatively healthy population. Three of these clustered in a single locus, 2q13 (110,86-110,98Mb), affecting the gene *NPHP1*. Homozygous deletions at this locus are known to cause the kidney disorder juvenile nephronophthisis. All three Biobank individuals with homozygous deletions at *NPHP1* had renal failure (Fisher exact test  $p = 9 \times 10^{-6}$ ). We also examined the data for the occurrence of two CNVs in the same person. 264 people carried two of these CNVs, not significantly different from the 249 expected by chance. All combinations of two CNVs observed in the same person are presented in Supplementary Table 14.

**Monitoring of CNV carriers.** Our results indicate a need for regular medical monitoring of apparently healthy carriers of specific pathogenic CNV. Examples include monitoring for blood pressure, kidney function and glucose levels for carriers of 16p12.1 and 16p11.2 deletions, and for cancer in 3q29 duplication carriers. Apart from specific medical phenotypes, it appears that such carriers require enhanced medical monitoring in general, as

their health can be affected in multiple ways. Our results should enable clinicians to better plan the medical management of CNV carriers.

Finally, the reported CNV morbidity map can provide researchers with another avenue for the elucidation of pathophysiological disease mechanisms.

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**Contributorship statement.** KC, MB-S and DO analysed the data, KMK, ER, and MB-S called the CNVs, AFP and ME contributed to the bioinformatics and website design, VE-P, JTRW, MCO'D and MJO contributed to the statistical analysis, JTRW, MCO'D and MJO edited the paper, GK conceived the project, drafted the paper and took part in all analysis steps.

## **Legends to Figures**

**Figure 1. Distribution of all 3132 p-values from CNV-phenotype associations.** There are 330 nominally significant CNV/phenotype associations ( $p < 0.05$ ), instead of the 157 expected by chance.

**Figure 2. ORs and 95% CI for the ORs for developing the 58 tested phenotypes in carriers of any one of the 49 rare pathogenic CNVs.** The phenotypes are ordered by the strength of the p-value.

**Figure 3. Relative Risk (RR) for dying during the follow-up to 2016 for carriers of the 54 CNVs.** The CNVs are ordered by the strength of the significance (strongest result on the left, for 16p13.11 duplications). The vertical line demarcates the nominally significant results ( $p < 0.05$ ). Due to zero observations in cases for some CNVs, RRs are shown, instead of ORs.

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## Supplementary material

### Medical consequences of pathogenic CNVs in adults: Analysis of the UK Biobank, by Crawford et al.

**Supplementary Table 1. List of CNVs analysed in the study.** “Genomic disorders (Cooper et al, 2011)” shows the list of CNVs proposed by Cooper et al,[8]; “Significant (Coe et al, 2014)” shows the CNVs that were significantly associated with neurodevelopmental disorders in Coe et al,[3]; “Genomic disorder (Dittwald et al, 2013)” shows the CNV list proposed by Dittwald et al.[6] “B-H FDR 0.1” indicates the number of phenotypes that were significantly associated with the CNV at a Benjamini-Hochberg FDR=0.1. The last two columns present known medical phenotypes associated with the CNVs analysed in the current study, and the literature sources they were derived from. We only considered large studies and do not include data on developmental delay, autism spectrum disorders, other neurodevelopmental disorders, behavioural problems or dysmorphisms. An “na” denotes an absence of medical phenotypes reported in the key papers, although other phenotypes were reported. The Numbers of carriers include first-degree relatives. The number of genes in the CNV are the numbers in the typical regions, indicated by the “Location” according to hg19.

CNV locus	Location (hg19)	N genes in CNV	N carriers	"Genomic disorders" (Cooper, 2011)	Significant (Coe, 2014)	"Genomic disorder" (Dittwald et al, 2013)	Unreliable	Rare	Include in analysis	B-H FDR <0.1	Medical Phenotypes (excluding developmental delay and neuropsychiatric phenotypes)	Source / reference
1p36del	chr1:0-2,500,000	76	1	X	X			X				
TAR_del	chr1:145,39-145,81	17	75	X	X	X			X	0	Limb anomalies, renal, cardiac, "Thrombocytopenia absent radius"	OMIM, Klopocki et al, 2007 (PMID:17236129); Rosenfeld et al, 2012 (PMC3376272)
TAR_dup	chr1:145,39-145,81	17	436	X	X	X			X	1	Skeletal/facial dysmorphism, ophthalmological	Rosenfeld et al, 2012 (PMC3376272)
1q21.1del	chr1:146,53-147,39	9	113	X	X	X			X	2	Short stature, cardiac, cataracts, microcephaly	OMIM; Brunetti-Pieri et al, 2008 (PMID:19029900)



1q21.1dup	chr1:146,53-147,39	9	177	X	X	X			X	1	Short stature, scoliosis, gastric uclers, macrocephaly	OMIM; Brunetti-Pieri et al, 2008 (PMID:19029900)
NRXN1del	chr2:50,14-51,26	1	163		X				X	1	Skeletal, cardiac, asthma/allergies	Dabel et al, 2013 (PMID:23495017)
2q11.2del	chr2:96,74-97,68	22	31	X	X	X			X	0	na	Riley et al, 2015 (PMID:26227573)
2q11.2dup	chr2:96,74-97,68	22	29	X		X			X	0	Short stature, gastrointestinal reflux	Riley et al, 2015 (PMID:26227573)
2q13del( <i>NPHP1</i> )	chr2:110,86-110,98	3	2448	X		X			X(common)	0	Nephronophthisis 1	OMIM
2q13dup( <i>NPHP1</i> )	chr2:110,86-110,98	3	1976	X		X			X(common)	0		-
2q13del	chr2:111,39-112,01	3	53		X	X			X	0	Cardiac, hypogonadism, hypospadias	Riley et al, 2015 (PMID:26227573)
2q13dup	chr2:111,39-112,01	3	71		X	X			X	1	na	Riley et al, 2015 (PMID:26227573)
2q21.1del	chr2:131,48-131,93	5	41			X			X	0	na	Dharmadhikari et al, 2012 (PMID:22543972)
2q21.1dup	chr2:131,48-131,93	5	59			X			X	0	na	Dharmadhikari et al, 2012 (PMID:22543972)
2q37del	chr2:239,72-243,2	51	1	X	X			X				
2q37dup	chr2:239,72-243,2	51	1	X				X				
3q29del	chr3:195,72-197,35	28	9	X	X	X			X	0	gastrointestinal, ear, dental, cardiac	Glassford et al, 2016 (PMID:26738761)
3q29dup	chr3:195,72-197,35	28	5	X		X			X	6	Cardiac	Tassano et al, 2018 (PMID:29501613)
W-Hdel	chr4:1,55-2,09	13	0	X	X			X				
W-Hdup	chr4:1,55-2,09	13	3	X	X			X				
Sotos_del	chr5:175,72-177,05	39	0	X	X	X		X				
5q35dup	chr5:175,72-177,05	39	0	X		X		X				
6q16del( <i>SIMI</i> )	chr6:100,84-100,91	1	5	X			X					
6q16dup( <i>SIMI</i> )	chr6:100,84-100,91	1	2	X			X	X				
WBS_del	chr7:72,74-74,14	26	1	X	X	X		X				
WBS_dup	chr7:72,74-74,14	26	14	X	X	X			X	0	Constipation, cardiac,	Morris et al, 2015 (PMID:26333794)
7q11.23del_distal	chr7:75,14-76,06	16	1	X		X		X				
7q11.23dup_distal	chr7:75,14-76,06	16	24	X		X			X	0	na	Foudes et al, 2016 (PMID:27867344)
8p23.1del	chr8:8,10-11,87	35	4	X	X	X		X				

8p23.1dup	chr8:8,10-11,87	35	6	X	X	X			X	0	Cardiac	Barber et al, 2013 (PMID:23345203)
EHMT1del	chr9:140,51-140,73	2	0					X				
EHMT1dup	chr9:140,51-140,73	2	1		X			X				
10q11.21q11.23del	chr10:49,39-51,06	19	57			X			X	0	consipation, GI reflux, epilepsy, sleep apnoea	Stankiewicz et al, 2012 (PMID:21948486)
10q11.21q11.23dup	chr10:49,39-51,06	19	43			X			X	0	na	Stankiewicz et al, 2012 (PMID:21948486)
10q23del	chr10:82,05-88,93	29	3	X	X	X		X				
10q23dup	chr10:82,05-88,93	29	7	X		X			X	0	na	van Bon et al, 2010 (PMID:21248748)
Potocki-Shaffer_del	chr11:43,94-46,02	22	0	X	X			X				
11p11.2dup	chr11:43,94-46,02	22	1	X				X				
13q12del( <i>CRYL1</i> )	chr13:20,98-21,10	2	379	X					X	0	na	DECIPHER
13q12dup( <i>CRYL1</i> )	chr13:20,98-21,10	2	10	X					X	0	na	DECIPHER
13q12.12del	chr13:23,56-24,88	10	85			X			X	0	Spastic ataxia, Charlevoix-Saguenay	OMIM
13q12.12dup	chr13:23,56-24,88	10	236			X			X	0	na	DECIPHER
15q11.2del	chr15:22,81-23,09	5	1664	X	X	X			X(common)	0	Neurological	OMIM, Cox & Butler, 2015 (PMC4346944)
15q11.2dup	chr15:22,81-23,09	5	2041	X	X	X			X(common)	0	na	OMIM
PWS/AS_del	chr15:23,68-28,39	116	1	X	X	X		X				
PWS/AS_dup	chr15:23,68-28,39	116	19	X	X	X			X	0	Dysmorphic, hypotonia, neurological	Piard et al, 2010 (PMID:20635369)
15q11q13del_BP3-BP4 ( <i>APBA2, TJP</i> )	chr15:29,16-30,38	4	16			X			X	1	seizures, hypotonia	Rosenfeld et al, 2011 (PMID:21248749)
15q11q13dup_BP3-BP4( <i>APBA2, TJP</i> )	chr15:29,16-30,38	4	53			X			X	0	na	DECIPHER
15q11q13del_BP3 BP5	chr15:29,16-32,46	17	1			X		X				
15q11q13dup_BP3 BP5	chr15:29,16-32,46	17	9			X			X	0	na	van Bon et al, 2009 (PMID:19372089)
15q13.3del	chr15:31,08-32,46	8	42	X	X	X			X	2	Seizures	Deutsch et al 2016 (PMID:26257138)
15q13.3dup	chr15:31,08-32,46	8	240	X		X			X	0	na	Hassfurth et al, 2016 (PMID:26997942)

15q13.3del( <i>CHRNA7</i> )	chr15:32,02-32,45	1	10			X			X	0	Seizures	Gillentine & Schaaf, 2015 (PMID:26095975)
15q13.3dup( <i>CHRNA7</i> )	chr15:32,02-32,45	1	3031			X			X(common)	0	na	Gillentine & Schaaf, 2015 (PMID:26095975)
15q24del	chr15:72,90-78,15	77	1	X	X	X		X				
15q24dup	chr15:72,90-78,15	77	9	X	X	X			X	0	Hearing loss, hernias	Nevado et al, 2014 (PMID:24764755)
15q25del	chr15:83,22-85,72	34	0	X	X	X		X				
15q25dup	chr15:83,22-85,72	34	3	X		X		X				
Rubinstein-Taybi_del	chr16:3,77-3,93	1	2	X				X				
Rubinstein-Taybi_dup	chr16:3,77-3,93	1	1	X				X				
16p13.11del	chr16:15,51-16,29	7	131	X	X	X			X	1	na	Watson et al, 2014 (PMID:24773319 )
16p13.11dup	chr16:15,51-16,29	7	828	X	X	X			X	2	na	Watson et al, 2014 (PMID:24773319 )
16p12.2-p11.2 del7.1-8.7-Mb	chr16:21,59-28,35	55	0	X		X		X				
16p12.2-p11.2 dup7.1-8.7-Mb	chr16:21,59-28,35	55	1	X		X		X				
16p12.1del	chr16:21,95-22,43	8	246	X	X	X			X	7	Skeletal, congenital cardiac, seizures, hypotonia, microcephaly	Girirajan et al, 2010 (PMID:20154674)
16p12.1dup	chr16:21,95-22,43	8	202	X		X			X	0	-	-
16p11.2distal_del	chr16:28,82-29,05	11	58	X	X	X			X	3	Obesity	Bachmann-Gagescu et al, 2010 (PMID:20808231)
16p11.2distal_dup	chr16:28,82-29,05	11	137	X	X	X			X	0	-	-
16p11.2del	chr16:29,65-30,20	30	110	X	X	X			X	7	Congenital abnormalities, seizures, eye accommodation, obesity	Shinawi et al, 2009 (PMID19914906); Jacquemont et al, 2011 (PMC3637175)
16p11.2dup	chr16:29,65-30,20	30	138	X	X	X			X	2	Congenital abnormalities, seizures, eye accommodation, obesity	Shinawi et al, 2009 (PMID19914906), Jacquemont et al, 2011 (PMC3637175)
17p13.3( <i>YWHA</i> E)del	chr17:1,25-1,30	1	26	X	X		X					
17p13.3( <i>YWHA</i> E)dup	chr17:1,25-1,30	1	8	X	X		X					
17p13.3( <i>PAFAH1B1</i> )del	chr17:2,49-2,59	1	1	X	X		X	X				
17p13.3( <i>PAFAH1B1</i> )dup	chr17:2,49-2,59	1	2	X	X		X	X				

17p12del(HNPP)	chr17:14,14-15,43	8	237	X		X		X	1	HNPP	Lupski et al, 1992 (PMID:1301995)
17p12dup(CMT1A)	chr17:14,14-15,43	8	124	X		X		X	3	CMT1A	Lupski et al, 1992 (PMID:1301995)
Smith Magenis Syndrome	chr17:16,81-20,21	59	2	X	X	X		X			
Potocki-Lupski Syndrome	chr17:16,81-20,21	59	5	X	X	X		X	0	Potoki-Lupski Syndrome	Potoki et al, 2007 (PMID:17357070)
17q11.2del( <i>NFI</i> )	chr17:29,12-30,27	19	9	X	X	X		X	0	Neurofibromatosis 1 (may have more tumours, cardiac and skeletal problems)	Kehrer-Sawatzki et al, 2017 (PMID:28213670)
17q11.2dup( <i>NFI</i> )	chr17:29,12-30,27	19	2	X	X	X		X			
17q12del	chr17:34,81-36,22	17	9	X	X	X		X	2	Kidney, liver, diabetes, ophthalmological, "Renal cysts and diabetes syndrome"	Rasmussen et al, 2016 (PMID:27409573)
17q12dup	chr17:34,81-36,22	17	101	X	X	X		X	1	Ophthalmological	Rasmussen et al, 2016 (PMID:27409573)
17q21.31del	chr17:43,70-44,29	10	0	X	X	X		X			
17q21.31dup	chr17:43,70-44,29	10	4	X		X		X			
17q23.1q23.2del	chr17:58,30-60,29	13	0			X		X			
17q23.1q23.2dup	chr17:58,30-60,29	13	2			X		X			
22q11.2del	chr22:19,04-21,47	61	10	X	X	X		X	0	VCFS/DGS, multiple phenotypes	OMIM; McDonald-McGinn et al, (PMID:27189754 )
22q11.2dup	chr22:19,04-21,47	61	280	X	X	X		X	2	Hypotonia, seizures, vision, hearing, heart defects	Wentzl et al, 2008 (PMID18707033)
22q11.2distal_del	chr22:21,92-23,65	26	5	X	X	X		X	1	Congenital, cardiac	Garavelli et al, 2011 (PMID:22582037); D'Angelo et al, 2018 (PMID:29441128)
22q11.2distal_dup	chr22:21,92-23,65	26	13	X	X	X		X	0	Seizures, facial dysmorphism, urogenital, hypotonia	Pinchefsky et al, 2017 (PMID:29147671)
SHANK3_del	chr22:51,11-51,17	1	0	X	X			X			
SHANK3_dup	chr22:51,11-51,17	1	0	X	X			X			

**Supplementary Table 2. CNV Calling Criteria.** Criteria used for calling CNVs (following our previous report, Kendall et al, 2016). CNVs at *EHMT1* and *SHANK3* were required to intersect at least 1Mbp distance, as small deletions and duplications were found to be common in samples with poor QC criteria, indicating that small CNVs in these telomeric regions were likely to be false-positives.

CNV	Criteria
1p36 del/dup	Size >50% of critical region, affecting <i>GABRD</i>
TAR del/dup	Size >50% of critical region
1q21.1 del/dup	Size >50% of critical region
<i>NRXN1</i> del	Exonic deletions
2q11.2 del/dup	Size >50% of critical region, affecting both <i>LMAN2L</i> and <i>ARID5A</i>
2q13 del/dup	Size >50% of critical region
2q13 del/dup ( <i>NPHP1</i> )	Size >50% of critical region, affecting <i>NPHP1</i>
2q21.1 del/dup	Size >50% of critical region
2q37 del/dup ( <i>HDAC4</i> )	Size >50% of critical region, affecting <i>HDAC4</i>
3q29 del/dup	Size >50% of critical region
Wolf-Hirschhorn del/dup	Size >50% of critical region
Sotos Syn/5q35 dup	Size >50% of critical region
6q16 del/dup ( <i>SIM1</i> )	Exonic deletions; whole gene duplications
Williams Beuren Syn del/dup	Size >50% of critical region
7q11.23 distal del/distal dup	Size >50% of critical region
8p23.1 del/dup	At least 1Mbp of critical region
9q34 del/dup ( <i>EHMT1</i> )	At least 1Mbp CNVs, including <i>EHMT1</i>
10q11.21q11.23 del/dup	Size >50% of critical region
10q23 del/dup	At least 1Mbp, including <i>NRG3</i> and <i>GRID1</i>
Potocki-Shaffer Syn del/11p11.2 dup ( <i>EXT2</i> )	Size >50% of critical region, including <i>EXT2</i>
13q12 del/dup ( <i>CRYL1</i> )	Exonic deletions; whole gene duplications
13q12.12 del/dup	Size >50% of critical region
15q11.2 del/dup	Size >50% of critical region
PWS del/dup	Full critical region, ~4Mbp
15q11q13 del/dup BP3-BP4	Size >50% of critical region
15q11q13 del/dup BP3-BP5	Size >50% of critical region
15q13.3 del/dup	Size >50% of critical region
15q13.3 del/dup ( <i>CHRNA7</i> )	Size >50% of critical region, affecting <i>CHRNA7</i>
15q24 del/dup	At least 1Mbp between the A-E intervals
15q25 del/dup	At least 1Mbp between the A-D intervals
Rubinstein-Taybi del/dup ( <i>CREBBP</i> )	Exonic deletions; whole gene duplications
16p13.11 del/dup	Size >50% of critical region

16p12.1 del/dup	Size >50% of critical region
16p12.2-p11.2 del/dup (7.1-8.7 Mb)	Size >50% of critical region
16p11.2 distal del/distal dup	Size >50% of critical region
16p11.2 del/dup	Size >50% of critical region
17p13.3 del/dup ( <i>YWHAE</i> )	Exonic deletions; whole gene duplications
17p13.3 del/dup ( <i>PAFAH1B1</i> )	Exonic deletions; whole gene duplications
17p12 del (HNPP)/dup (CMT1A)	Size >50% of critical region, affecting <i>PMP22</i>
Smith-Magenis/Potocki-Lupski Syn	Size >50% of critical region
17q11.2 del/dup ( <i>NFI</i> )	Size >50% of critical region, affecting <i>NFI</i>
17q12 del/dup	Size >50% of critical region
17q21.31 del/dup	Size >50% of critical region
17q23.1q23.2 del/dup	Size >50% of critical region
22q11.2 del/dup	Size >50% of critical region
22q11.2 distal del/dup	Size >50% of critical region
<i>SHANK3</i> del/dup	At least 1Mbp CNVs, including <i>SHANK3</i>
“Large” CNVs	Size > 20Mbp + >50 genes

**Supplementary Table 3. List of UK Biobank disease codes used to construct each phenotype group.**

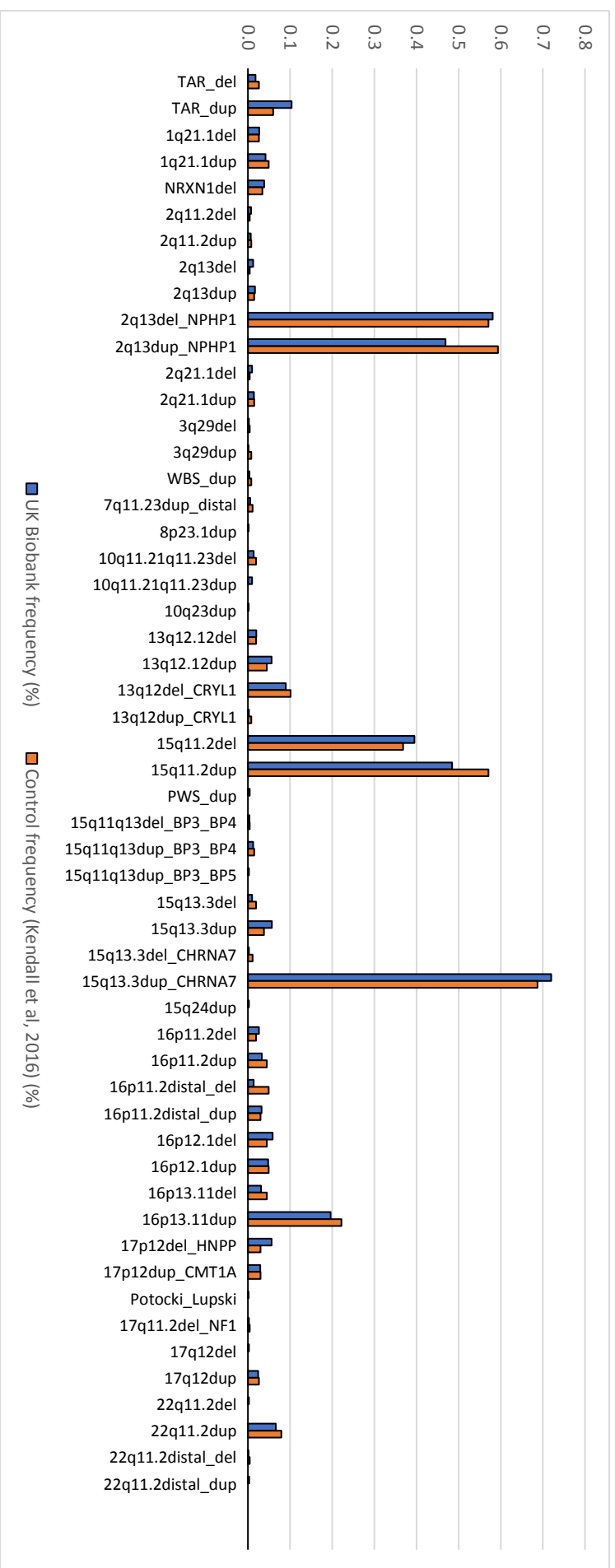
(This table is available as a stand-alone file)

**Supplementary Table 4. Comparison of CNV frequencies with previous control populations.** The frequencies of the 54 CNV in the UK Biobank (including first-degree relatives, but excluding people who are not white British or Irish) are compared with 26,626 population controls from previous studies reported by us. These other controls originated from various datasets where we had access to the raw data, or had completed all the analysis ourselves, applying the same criteria. Details on these control datasets are presented in our previous publication [10].

CNV	Number of carriers in UK Biobank	UK Biobank frequency (%)	Control frequency (Kendall et al, 2016) (%)
TAR_del	75	0.018	0.026
TAR_dup	436	0.103	0.060
1q21.1del	113	0.027	0.026
1q21.1dup	177	0.042	0.049
NRXN1del	163	0.039	0.034
2q11.2del	31	0.007	0.004
2q11.2dup	29	0.007	0.008
2q13del	53	0.013	0.004
2q13dup	71	0.017	0.015
2q13del_NPHP1	2448	0.581	0.571
2q13dup_NPHP1	1976	0.469	0.593
2q21.1del	41	0.010	0.004
2q21.1dup	59	0.014	0.015
3q29del	9	0.002	0.004
3q29dup	5	0.001	0.008
WBS_dup	14	0.003	0.008
7q11.23dup_distal	24	0.006	0.011
8p23.1dup	6	0.001	0.000
10q11.21q11.23del	57	0.014	0.019
10q11.21q11.23dup	41	0.010	0.000
10q23dup	7	0.002	0.000
13q12.12del	85	0.020	0.019
13q12.12dup	236	0.056	0.045
13q12del_CRYL1	379	0.090	0.101
13q12dup_CRYL1	10	0.002	0.008
15q11.2del	1664	0.395	0.368
15q11.2dup	2041	0.484	0.571
PWS_dup	19	0.005	0.000
15q11q13del_BP3_BP4	16	0.004	0.004
15q11q13dup_BP3_BP4	53	0.013	0.015
15q11q13dup_BP3_BP5	9	0.002	0.000
15q13.3del	42	0.010	0.019
15q13.3dup	240	0.057	0.038
15q13.3del_CHRNA7	10	0.002	0.011
15q13.3dup_CHRNA7	3031	0.719	0.687
15q24dup	9	0.002	0.000

16p11.2del	110	0.026	0.019
16p11.2dup	138	0.033	0.045
16p11.2distal_del	58	0.014	0.049
16p11.2distal_dup	137	0.033	0.030
16p12.1del	246	0.058	0.045
16p12.1dup	202	0.048	0.049
16p13.11del	131	0.031	0.045
16p13.11dup	828	0.197	0.222
17p12del HNPP	237	0.056	0.030
17p12dup CMT1A	124	0.029	0.030
Potocki Lupski	5	0.001	0.000
17q11.2del NF1	9	0.002	0.004
17q12del	9	0.002	0.000
17q12dup	101	0.024	0.026
22q11.2del	10	0.002	0.000
22q11.2dup	280	0.066	0.079
22q11.2distal_del	5	0.001	0.004
22q11.2distal_dup	13	0.003	0.000





**Supplementary Figure 1. Comparison of CNV frequencies with previous control populations.** The figure is based on Supplementary Table 4, above.

**Supplementary Table 5. CNV calls in each batch.** The UK Biobank genotyped the samples in 106 batches. This table presents the numbers of each CNV in each of these batches, for white British or Irish subjects, including first-degree relatives, and after array QC. There are no outliers from the expected Poisson distribution, after multiple testing correction, indicating a lack of batch effects on the calling of these 54 CNVs.

(This table is available as a stand-alone file)

**Supplementary Table 6. All CNV/phenotype associations, grouped by CNV.** The nomenclature follows that for Table 1 in the main text.

(This table is available as a stand-alone file)

**Supplementary Table 7. All CNV/phenotype associations, grouped by phenotype.** Significant results at FDR=0.1 are shown in bold.

(This table is available as a stand-alone file)

**Supplementary Table 8. Causes of death during follow-up among CNV carriers.** The list is sorted by CNV.

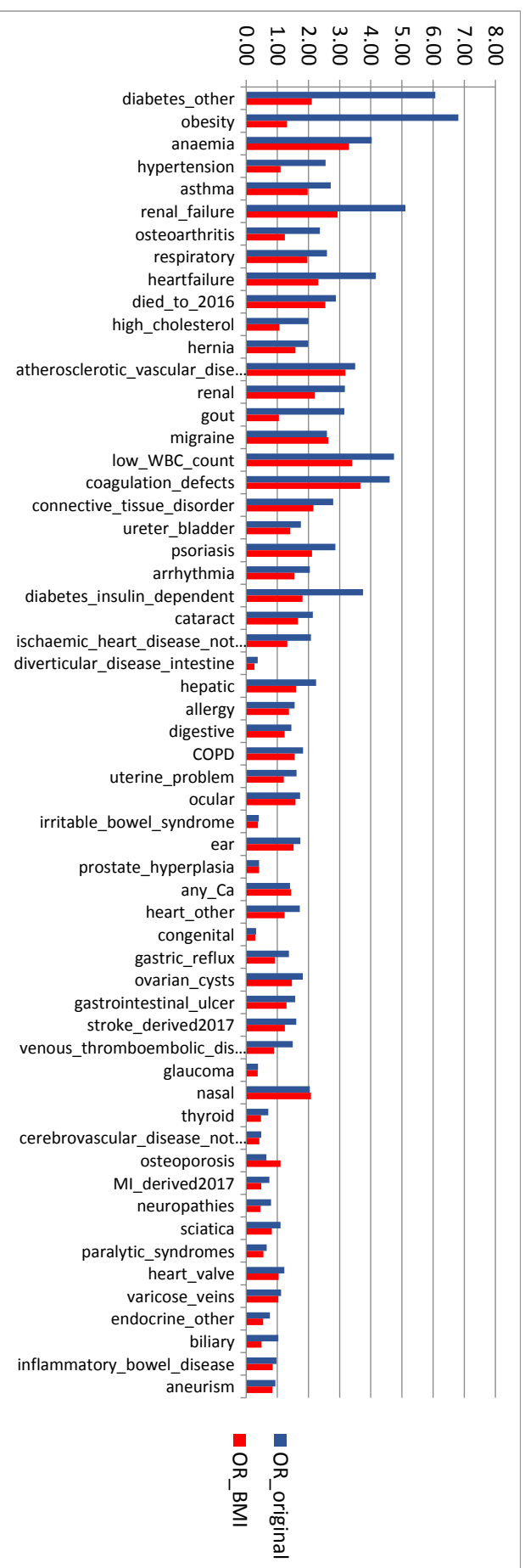
(This table is available as a stand-alone file)

**Supplementary Table 9. Effect of 16p11.2 deletions on phenotypes before and after correction with BMI.** Obesity is a known consequence of deletions at 16p11.2. We tested whether obesity is the factor leading to other (“novel”) associations. The table shows the changes to p-values and ORs after BMI was added as a co-variate to Firth’s logistic regression analysis. Other co-variables were kept the same. Significant results at FDR=0.1 are shown in red. Excluding obesity, four of six originally significant associations are still significant.

Phenotype	p-value original	p-value BMI	B-H FDR	OR original	OR BMI	OR Lower 95% original	OR Upper 95%_original	OR Lower 95%_BMI	OR Upper 95%_BMI
diabetes_other	2.54E-11	0.0064	0.088	6.07	2.11	3.77	9.50	1.24	3.48
obesity	7.39E-10	0.43	0.76	6.81	1.31	4.01	11.01	0.66	2.48
anaemia	2.15E-06	5.40E-05	0.0050	4.03	3.30	2.38	6.48	1.92	5.36
hypertension	9.44E-06	0.66	0.90	2.55	1.11	1.69	3.83	0.71	1.72
asthma	1.33E-05	0.0030	0.052	2.71	1.97	1.77	4.08	1.27	3.00
renal_failure	6.04E-05	0.0051	0.078	5.11	2.93	2.49	9.45	1.42	5.47
osteoarthritis	0.00031	0.36	0.70	2.36	1.24	1.50	3.63	0.78	1.95
respiratory	0.0023	0.026	0.19	2.59	1.97	1.44	4.37	1.09	3.33
heartfailure	0.0045	0.074	0.36	4.16	2.31	1.64	8.84	0.91	4.93
died_to_2016	0.0050	0.012	0.11	2.88	2.54	1.42	5.27	1.25	4.66
high_cholesterol	0.0058	0.80	0.96	2.00	1.06	1.23	3.15	0.65	1.70
hernia	0.0084	0.072	0.36	1.99	1.58	1.20	3.16	0.96	2.52
atherosclerotic_vascular_disease	0.010	0.016	0.13	3.50	3.19	1.40	7.33	1.27	6.67
renal	0.015	0.083	0.38	3.16	2.20	1.28	6.49	0.89	4.54
gout	0.017	0.92	1	3.15	1.05	1.26	6.64	0.37	2.42
migraine	0.020	0.019	0.15	2.59	2.63	1.18	4.98	1.20	5.06
low_WBC_count	0.021	0.11	0.39	4.74	3.41	1.32	11.90	0.71	9.78
coagulation_defects	0.024	0.048	0.29	4.60	3.67	1.28	11.58	1.01	9.26
connective_tissue_disorder	0.029	0.091	0.39	2.79	2.16	1.13	5.76	0.87	4.47
ureter_bladder	0.034	0.18	0.50	1.76	1.41	1.05	2.81	0.84	2.27
psoriasis	0.039	0.13	0.43	2.86	2.11	1.06	6.16	0.78	4.58
arrhythmia	0.040	0.20	0.51	2.05	1.54	1.04	3.68	0.78	2.78
diabetes_insulin_dependent	0.045	0.33	0.67	3.75	1.81	1.04	9.48	0.49	4.70
cataract	0.064	0.23	0.54	2.14	1.66	0.95	4.23	0.70	3.38
ischaemic_heart_disease_not_MI	0.071	0.47	0.11	2.08	1.32	0.93	4.06	0.59	2.58
diverticular_disease_intestine	0.071	0.010	0.79	0.37	0.26	0.08	1.08	0.05	0.77
hepatic	0.077	0.28	0.62	2.24	1.60	0.91	4.61	0.65	3.32
allergy	0.081	0.21	0.53	1.56	1.37	0.94	2.46	0.83	2.18
digestive	0.099	0.36	0.70	1.45	1.23	0.93	2.20	0.79	1.87
COPD	0.12	0.24	0.55	1.83	1.55	0.85	3.52	0.72	2.99
uterine_problem	0.14	0.57	0.87	1.62	1.20	0.84	2.95	0.63	2.21
ocular	0.16	0.23	0.54	1.73	1.58	0.79	3.31	0.72	3.03
irritable_bowel_syndrome	0.20	0.17	0.49	0.41	0.38	0.05	1.47	0.04	1.39
ear	0.21	0.33	0.67	1.74	1.52	0.70	3.56	0.62	3.12
prostate_hyperplasia	0.22	0.23	0.56	0.41	0.42	0.05	1.55	0.05	1.57
any_Ca	0.26	0.22	0.55	1.41	1.45	0.77	2.40	0.79	2.47
heart_other	0.26	0.65	0.89	1.72	1.23	0.63	3.75	0.45	2.69
congenital	0.31	0.27	0.61	0.31	0.29	0.00	2.15	0.00	1.97
gastric_reflux	0.32	0.81	0.83	1.37	0.93	0.72	2.39	0.49	1.64
ovarian_cysts	0.32	0.51	0.96	1.82	1.47	0.50	4.71	0.40	3.83
gastrointestinal_ulcer	0.34	0.57	0.88	1.57	1.30	0.58	3.44	0.48	2.84
stroke_derived2017	0.37	0.67	0.91	1.61	1.24	0.52	3.76	0.41	2.90

venous thromboembolic disease	0.39	0.79	0.96	1.49	0.89	0.55	3.22	0.33	1.94
glaucoma	0.41	0.40	0.74	0.37	0.37	0.00	2.59	0.00	2.56
nasal	0.44	0.43	0.78	2.05	2.08	0.23	7.42	0.24	7.55
thyroid	0.47	0.093	0.38	0.71	0.48	0.23	1.66	0.16	1.12
cerebrovascular_disease_not_stroke	0.56	0.48	0.80	0.48	0.42	0.00	3.32	0.00	2.93
osteoporosis	0.58	0.91	0.47	0.65	1.10	0.07	2.42	0.12	4.14
MI_derived2017	0.58	0.15	0.99	0.74	0.48	0.20	1.93	0.13	1.25
neuropathies	0.67	0.11	0.41	0.80	0.46	0.22	2.00	0.13	1.17
sciatica	0.74	0.51	0.84	1.10	0.83	0.59	1.89	0.44	1.42
paralytic_syndromes	0.75	0.65	0.91	0.66	0.56	0.01	4.52	0.00	3.85
heart_valve	0.76	0.95	1	1.23	1.04	0.25	3.53	0.22	3.01
varicose_veins	0.84	0.96	0.92	1.12	1.03	0.31	2.81	0.29	2.58
endocrine_other	0.84	0.63	0.98	0.77	0.54	0.01	5.26	0.00	3.74
biliary	0.94	0.087	0.39	1.03	0.50	0.38	2.24	0.18	1.10
inflammatory_bowel_disease	0.95	0.71	0.93	0.97	0.85	0.36	2.09	0.32	1.83
aneurism	0.96	0.90	0.99	0.94	0.84	0.01	6.52	0.01	5.83

**Supplementary Figure 2. Effect of 16p11.2del on phenotypes before and after correction for BMI, based on the above table.** It shows the changes in ORs after correction for BMI. The CNVs are ordered according to the strength of the original p-value (strongest on the left). The first 23 CNVs were nominally significant ( $p < 0.05$ ) in the original analysis. The correction with BMI removes most of the effect on diabetes type 2, hypertension, heart and renal failure, while those on anaemia, atherosclerotic valve disease and mortality remain largely unchanged.

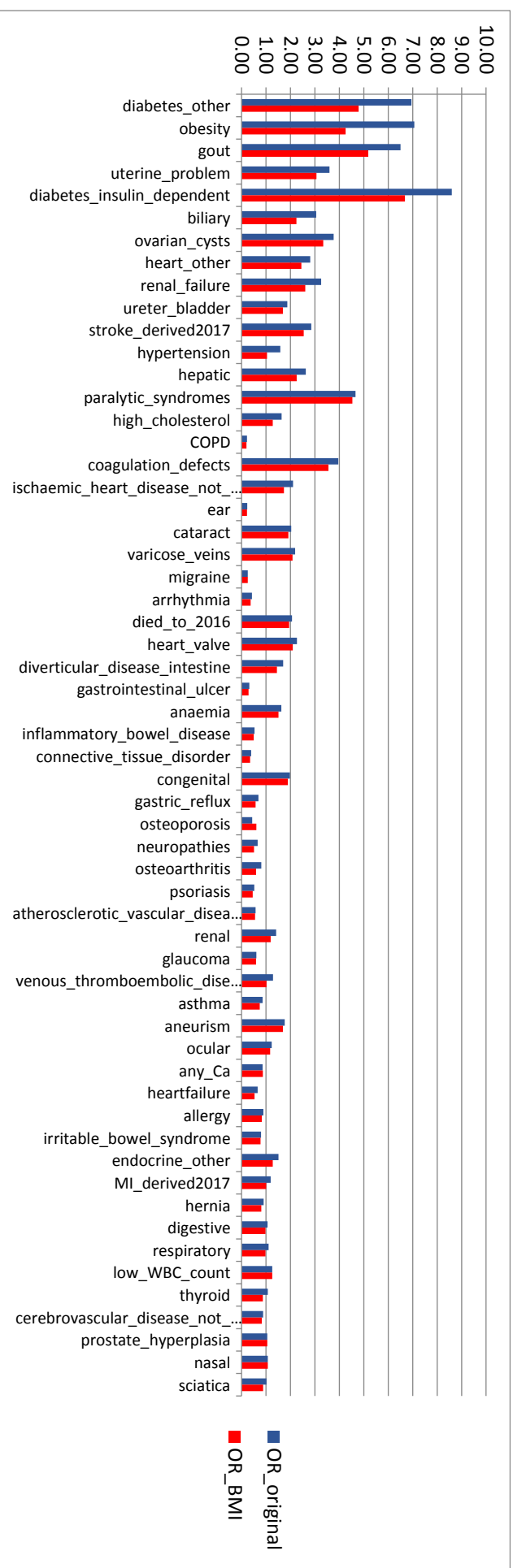


**Supplementary Table 10. Effect of “distal” 16p11.2 deletions on phenotypes before and after correction with BMI.** Obesity is a known consequence of this deletion. We tested whether obesity is the factor leading to other (“novel”) associations. The table shows the changes to p-values and ORs after BMI was added as a co-variate to Firth’s logistic regression analysis. Other co-variables were kept the same. Significant results at FDR=0.1 are shown in red. Excluding obesity, four observations are significant after correction for BMI.

Phenotype	p-value original	p-value BMI	B-H FDR	OR original	OR BMI	OR Lower 95% original	OR Upper 95%_original	OR Lower 95%_BMI	OR Upper 95%_BMI
diabetes_other	8.86E-08	1.39E-05	0.0038	6.95	4.79	3.67	12.57	2.47	8.89
obesity	1.14E-05	0.0015	0.038	7.06	4.25	3.27	13.73	1.81	9.11
gout	0.00046	0.0017	0.036	6.50	5.19	2.52	14.40	2.00	11.54
uterine_problem	0.0020	0.0067	0.084	3.60	3.06	1.63	7.97	1.38	6.82
diabetes insulin dependent	0.0033	0.0076	0.091	8.60	6.69	2.35	22.23	1.82	17.38
biliary	0.014	0.068	0.35	3.06	2.25	1.29	6.27	0.94	4.69
ovarian_cysts	0.050	0.071	0.36	3.77	3.34	1.00	10.35	0.89	9.21
heart_other	0.070	0.11	0.39	2.81	2.44	0.91	6.72	0.79	5.84
renal_failure	0.071	0.13	0.44	3.26	2.60	0.89	8.48	0.71	6.81
ureter_bladder	0.077	0.14	0.44	1.88	1.69	0.93	3.50	0.84	3.15
stroke_derived2017	0.10	0.14	0.45	2.86	2.54	0.78	7.41	0.69	6.61
hypertension	0.11	0.90	0.99	1.59	1.04	0.90	2.78	0.58	1.84
hepatic	0.13	0.19	0.50	2.63	2.26	0.72	6.77	0.62	5.82
paralytic_syndromes	0.14	0.14	0.45	4.66	4.54	0.53	17.22	0.51	16.79
high_cholesterol	0.15	0.47	0.80	1.64	1.28	0.83	3.05	0.64	2.38
COPD	0.16	0.13	0.45	0.22	0.20	0.00	1.54	0.00	1.42
coagulation_defects	0.17	0.20	0.52	3.96	3.55	0.45	14.57	0.40	13.11
ischaemic_heart_disease_not_MI	0.18	0.30	0.49	2.11	1.74	0.68	5.06	0.56	4.18
ear	0.18	0.16	0.64	0.23	0.22	0.00	1.62	0.00	1.51
cataract	0.20	0.23	0.55	2.03	1.92	0.65	4.92	0.62	4.64
varicose_veins	0.20	0.23	0.55	2.19	2.09	0.60	5.64	0.57	5.38
migraine	0.22	0.22	0.54	0.25	0.26	0.00	1.78	0.00	1.79
arrhythmia	0.24	0.17	0.50	0.42	0.37	0.05	1.57	0.04	1.39
died_to_2016	0.24	0.27	0.61	2.06	1.95	0.57	5.32	0.53	5.03
heart_valve	0.26	0.31	0.65	2.26	2.10	0.47	6.66	0.43	6.16
diverticular_disease_intestine	0.28	0.44	0.76	1.70	1.44	0.62	3.80	0.52	3.24
gastrointestinal_ulcer	0.32	0.27	0.62	0.31	0.29	0.00	2.19	0.00	2.00
anaemia	0.36	0.43	0.77	1.63	1.51	0.53	3.85	0.49	3.59
inflammatory_bowel_disease	0.39	0.34	0.68	0.53	0.49	0.06	1.94	0.06	1.82
connective_tissue_disorder	0.45	0.37	0.71	0.40	0.35	0.00	2.77	0.00	2.43
congenital	0.46	0.48	0.80	1.98	1.89	0.22	7.27	0.21	6.97
gastric_reflux	0.48	0.28	0.62	0.69	0.57	0.19	1.78	0.16	1.48
osteoporosis	0.50	0.70	0.92	0.44	0.61	0.00	3.10	0.00	4.32
neuropathies	0.58	0.36	0.71	0.65	0.51	0.07	2.41	0.06	1.89
osteoarthritis	0.59	0.18	0.52	0.81	0.60	0.34	1.67	0.25	1.25
psoriasis	0.61	0.52	0.84	0.52	0.45	0.00	3.63	0.00	3.16
atherosclerotic_vascular_disease	0.67	0.64	0.91	0.58	0.55	0.00	4.03	0.00	3.86
renal	0.69	0.84	0.96	1.41	1.19	0.16	5.19	0.14	4.41
glaucoma	0.70	0.69	0.92	0.60	0.60	0.00	4.24	0.00	4.19
venous_thromboembolic_disease	0.71	0.97	0.79	1.28	1.02	0.26	3.76	0.21	3.01
asthma	0.71	0.46	0.95	0.86	0.74	0.34	1.82	0.29	1.57
aneurism	0.71	0.73	0.99	1.77	1.69	0.01	12.50	0.01	11.92

ocular	0.72	0.78	0.95	1.23	1.17	0.34	3.16	0.32	3.02
any_Ca	0.74	0.75	0.95	0.86	0.87	0.31	1.92	0.32	1.94
heartfailure	0.76	0.62	0.91	0.66	0.53	0.01	4.67	0.00	3.76
allergy	0.76	0.62	0.92	0.89	0.83	0.38	1.81	0.35	1.69
irritable_bowel_syndrome	0.78	0.75	0.96	0.80	0.77	0.09	2.97	0.09	2.87
endocrine_other	0.79	0.87	0.98	1.51	1.28	0.01	10.52	0.01	8.94
MI_derived2017	0.80	0.99	0.91	1.19	1.01	0.24	3.57	0.21	3.02
hernia	0.80	0.61	0.99	0.90	0.81	0.36	1.93	0.32	1.73
digestive	0.84	0.95	1	1.07	0.98	0.54	1.95	0.50	1.80
respiratory	0.85	0.97	0.99	1.11	0.98	0.30	2.86	0.27	2.52
low_WBC_count	0.88	0.88	0.99	1.25	1.26	0.01	8.67	0.01	8.72
thyroid	0.90	0.80	0.96	1.07	0.87	0.29	2.81	0.23	2.30
cerebrovascular_disease_not_stroke	0.93	0.89	0.99	0.88	0.83	0.01	6.17	0.01	5.78
prostate_hyperplasia	0.94	0.94	1	1.05	1.05	0.21	3.28	0.21	3.28
nasal	0.96	0.96	0.99	1.07	1.07	0.01	7.48	0.01	7.45
sciatica	0.97	0.76	0.95	1.02	0.88	0.40	2.15	0.35	1.87

**Supplementary Figure 3. Effect of “distal” 16p11.2 deletions on phenotypes before and after correction for BMI, based on the above table.** The CNVs are ordered according to the strength of the original p-value (strongest on the left). The first six phenotypes on the left were nominally significant in the original analysis. A partial effect of BMI on the ORs can be seen on diabetes type 1 and 2, and gout.



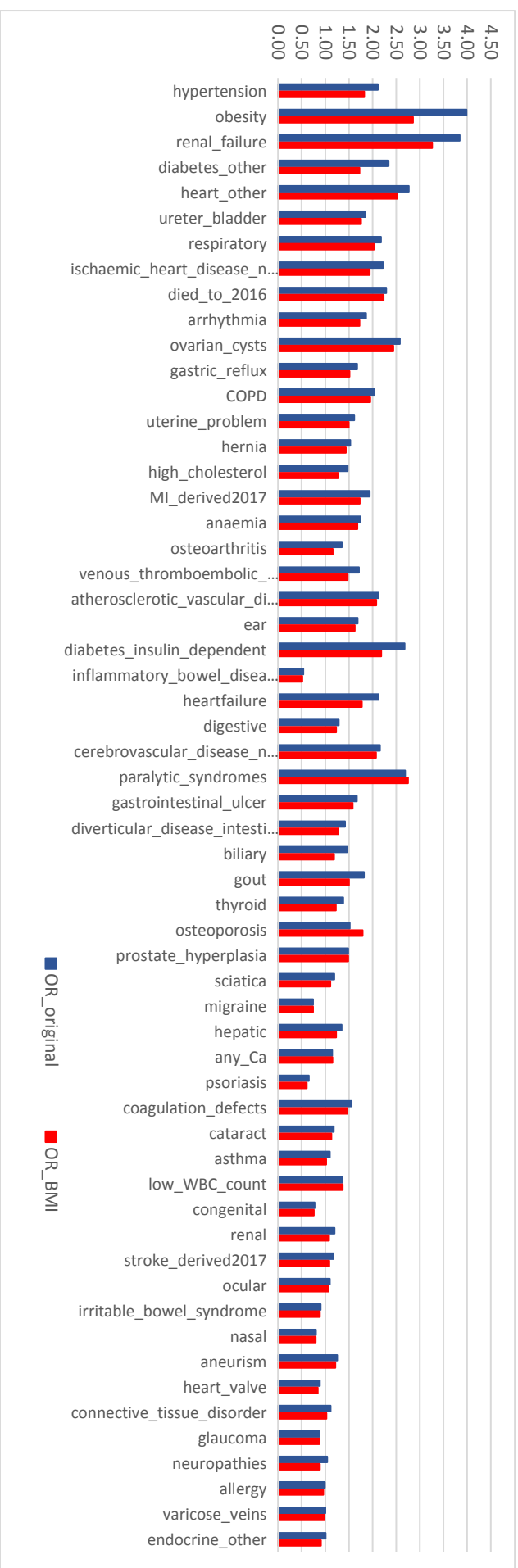


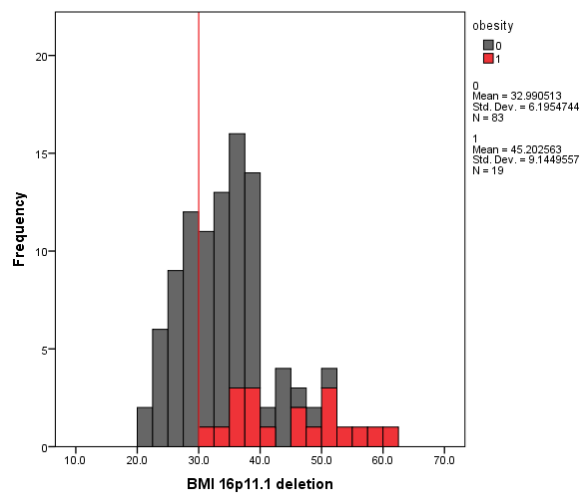
**Supplementary Table 11. Effect of 16p12.1 deletions on phenotypes before and after correction for BMI.** Obesity is not an established consequence of this deletion, but was a prominent finding in our analysis. We tested whether obesity is the factor leading to other (“novel”) associations. The table shows the changes to p-values and ORs after BMI was added as a co-variate to Firth’s logistic regression analysis. Other co-variables were kept the same. Significant results at FDR=0.1 are shown in red. Five of the original six associations (obesity excluded) remain significant and risk of death has also become significant.

Phenotype	p-value original	p-value BMI	B-H FDR	OR original	OR BMI	OR Lower 95% original	OR Upper 95%_original	OR Lower 95%_BMI	OR Upper 95%_BMI
hypertension	8.64E-08	4.02E-05	0.0055	2.11	1.83	1.61	2.76	1.37	2.43
obesity	1.11E-07	0.00027	0.011	3.99	2.85	2.53	5.99	1.66	4.66
renal failure	7.23E-06	6.99E-05	0.0039	3.85	3.26	2.26	6.13	1.90	5.25
diabetes_other	0.00021	0.023	0.17	2.34	1.72	1.53	3.46	1.08	2.64
heart_other	0.00029	0.00090	0.031	2.77	2.52	1.65	4.36	1.50	3.98
ureter_bladder	0.00033	0.0010	0.031	1.85	1.76	1.34	2.52	1.26	2.40
respiratory	0.00069	0.0019	0.037	2.18	2.03	1.42	3.21	1.32	3.00
ischaemic_heart_disease_not_MI	0.0023	0.011	0.10	2.22	1.94	1.35	3.45	1.18	3.03
died to 2016	0.0052	0.0066	0.087	2.29	2.23	1.31	3.72	1.27	3.62
arrhythmia	0.0069	0.017	0.14	1.86	1.72	1.20	2.78	1.11	2.57
ovarian_cysts	0.0070	0.010	0.11	2.58	2.44	1.33	4.52	1.26	4.29
gastric_reflux	0.0098	0.037	0.24	1.67	1.51	1.14	2.38	1.03	2.16
COPD	0.010	0.016	0.14	2.04	1.95	1.20	3.27	1.14	3.12
uterine_problem	0.016	0.043	0.27	1.61	1.49	1.10	2.33	1.01	2.16
hernia	0.020	0.046	0.28	1.53	1.44	1.07	2.13	1.01	2.00
high_cholesterol	0.022	0.16	0.48	1.47	1.27	1.06	2.02	0.91	1.76
MI_derived2017	0.025	0.061	0.34	1.94	1.73	1.09	3.19	0.97	2.86
anaemia	0.027	0.037	0.25	1.74	1.68	1.07	2.68	1.03	2.58
osteoarthritis	0.066	0.39	0.73	1.35	1.16	0.98	1.84	0.83	1.59
venous_thromboembolic_disease	0.068	0.18	0.50	1.71	1.47	0.96	2.81	0.82	2.44
atherosclerotic_vascular_disease	0.070	0.079	0.37	2.13	2.08	0.93	4.13	0.91	4.03
ear	0.075	0.096	0.38	1.69	1.63	0.95	2.77	0.91	2.67
diabetes_insulin_dependent	0.075	0.15	0.47	2.68	2.18	0.89	6.06	0.72	4.99
inflammatory_bowel_disease	0.081	0.064	0.35	0.53	0.52	0.22	1.07	0.21	1.03
heartfailure	0.092	0.19	0.50	2.13	1.77	0.87	4.33	0.72	3.64
digestive	0.10	0.18	0.51	1.28	1.23	0.95	1.71	0.91	1.64
cerebrovascular_disease_not_stroke	0.11	0.13	0.40	2.16	2.07	0.81	4.59	0.78	4.40
paralytic_syndromes	0.11	0.11	0.44	2.69	2.75	0.75	6.66	0.77	6.82
gastrointestinal_ulcer	0.14	0.18	0.51	1.67	1.58	0.84	2.95	0.79	2.80
diverticular_disease_intestine	0.14	0.30	0.64	1.42	1.28	0.89	2.16	0.80	1.95
biliary	0.14	0.50	0.83	1.46	1.19	0.88	2.29	0.70	1.89
gout	0.15	0.32	0.66	1.82	1.50	0.79	3.56	0.65	2.99
thyroid	0.17	0.38	0.72	1.38	1.23	0.86	2.10	0.76	1.88
osteoporosis	0.22	0.097	0.38	1.52	1.79	0.76	2.72	0.89	3.22
prostate_hyperplasia	0.24	0.24	0.56	1.49	1.49	0.75	2.70	0.75	2.70
sciatica	0.37	0.61	0.91	1.19	1.11	0.80	1.71	0.74	1.59
migraine	0.43	0.44	0.77	0.74	0.74	0.30	1.49	0.30	1.50
hepatic	0.44	0.59	0.89	1.35	1.23	0.59	2.60	0.54	2.37
any_Ca	0.47	0.46	0.78	1.15	1.15	0.78	1.64	0.78	1.65

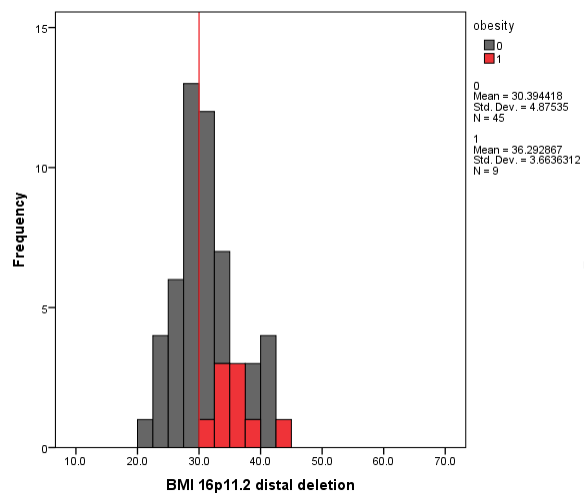
psoriasis	0.48	0.39	0.73	0.66	0.61	0.14	1.85	0.13	1.72
coagulation_defects	0.52	0.57	0.88	1.55	1.47	0.33	4.40	0.31	4.15
cataract	0.55	0.65	0.90	1.18	1.13	0.67	1.93	0.64	1.86
asthma	0.63	0.93	0.99	1.10	1.02	0.75	1.56	0.69	1.45
low_WBC_count	0.64	0.64	0.90	1.36	1.37	0.29	3.85	0.29	3.87
congenital	0.67	0.65	0.90	0.77	0.76	0.16	2.18	0.16	2.15
renal	0.69	0.86	0.97	1.20	1.08	0.45	2.53	0.40	2.28
stroke_derived2017	0.69	0.84	0.98	1.17	1.09	0.48	2.37	0.44	2.20
ocular	0.74	0.81	0.96	1.10	1.07	0.62	1.80	0.60	1.76
irritable_bowel_syndrome	0.77	0.74	0.95	0.90	0.89	0.40	1.74	0.39	1.71
nasal	0.78	0.78	0.95	0.80	0.80	0.09	2.87	0.09	2.86
aneurism	0.79	0.82	0.92	1.25	1.21	0.14	4.52	0.14	4.37
heart_valve	0.79	0.71	0.96	0.88	0.84	0.29	2.00	0.28	1.90
connective_tissue_disorder	0.79	0.95	1	1.11	1.03	0.46	2.24	0.42	2.07
glaucoma	0.81	0.80	0.96	0.88	0.88	0.25	2.19	0.24	2.18
neuropathies	0.90	0.70	0.92	1.04	0.88	0.52	1.83	0.44	1.57
allergy	0.97	0.82	0.96	0.99	0.96	0.68	1.40	0.66	1.35
varicose_veins	0.99	0.96	0.99	1.00	0.98	0.44	1.93	0.43	1.89
endocrine_other	0.99	0.90	1	1.01	0.91	0.12	3.61	0.10	3.26

**Supplementary Figure 4. Effect of 16p12.1 deletions on phenotypes before and after correction for BMI, based on the above table. BMI correction has little effect on the associated phenotypes.**

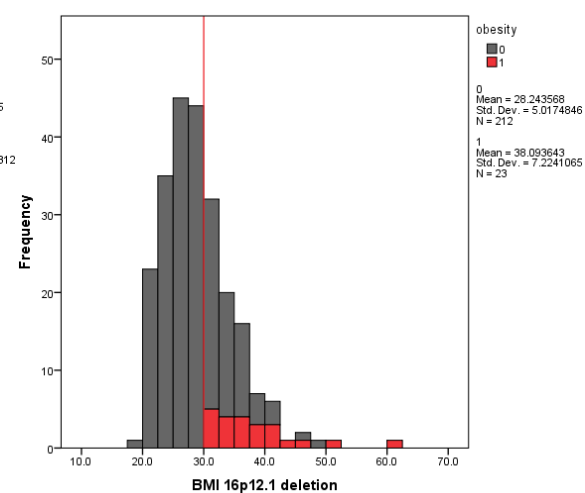




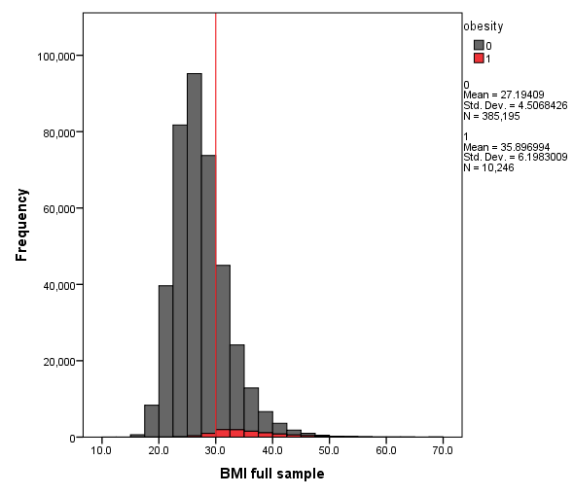
a



b



c



d

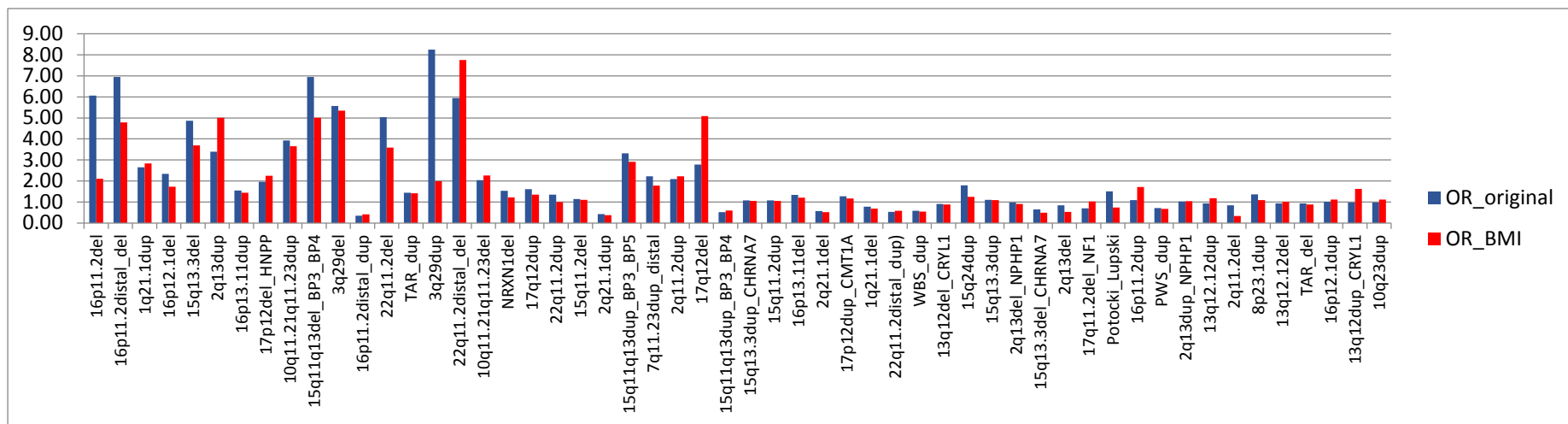
**Supplementary Figure 5 (a-d). Distribution of BMI in the CNVs associated with obesity.** The distribution of BMI is different in the three analysed CNVs. It is most strongly shifted towards high values in 16p11.2 deletions (a), where 71.6% of carriers have BMI>30. It is less shifted for 16p11.2 distal deletions (b), where 55.6% have a BMI>30, while in 16p12.1 deletion carriers (c), only 37% of carriers have BMI>30, making the distribution much closer to that in the full sample (d). The vertical red lines indicate the point where obesity could be diagnosed using the BMI>30 criterion. The carriers who had been diagnosed with an ICD10 hospital discharge code for “Obesity” (marked in red) also constitute correspondingly smaller proportions: 18.6%, 16.7% and 9.8% respectively for the three CNVs. Only 2.6% of the whole Biobank population had a diagnosis of Obesity, but 24.3% could be diagnosed with the BMI>30 criterion. i.e. only one in 10 people meeting the criterion received the medical diagnosis. These differences could be one reason why correcting for BMI causes different changes to the associations with obesity for the studied CNVs. From a purely statistical point of view, categorical and continuous traits (even if the category is derived from a threshold imposed on exactly the same measurement representing the continuous trait) are not equivalent, and therefore adjusting an analysis of one for another does not necessarily remove evidence for association (Obesity is a binary state, while BMI is a continuous one). These factors could explain why correcting for BMI does not completely remove the association with Obesity.

**Supplementary Table 12. Effect of all CNVs on type 2 diabetes before and after correction with BMI.** The ORs are reduced substantially for 16p11.2del, 16p11.2distal\_del and 3q29dup, suggesting that the diabetes in these CNV carriers is caused mostly via an increased BMI. For 1q21.1dup, 16p12.1del and 3q29del the ORs are unchanged, while for 2q13dup, 22q11.2distal\_del, and 17q12del they increase, suggesting that these CNVs have a more direct effect on the development of diabetes. Significant results at FDR=0.1 are shown in red, with six associations significant after correction for BMI.

Phenotype	p-value original	p-value BMI	B-H FDR	OR original	OR BMI	OR Lower 95% original	OR Upper 95%_original	OR Lower 95%_BMI	OR Upper 95%_BMI
16p11.2del	2.54E-11	0.0064	0.088	6.07	2.11	3.77	9.50	1.24	3.48
16p11.2distal_del	8.86E-08	1.39E-05	0.0038	6.95	4.79	3.67	12.57	2.47	8.89
1q21.1dup	0.00017	9.67E-05	0.0044	2.65	2.83	1.64	4.10	1.73	4.44
16p12.1del	0.00021	0.023	0.17	2.34	1.72	1.53	3.46	1.08	2.64
15q13.3del	0.00038	0.0059	0.086	4.87	3.69	2.15	10.15	1.50	8.30
2q13dup	0.0012	5.42E-05	0.0037	3.39	5.01	1.68	6.29	2.45	9.47
16p13.11dup	0.0018	0.010	0.10	1.55	1.45	1.18	1.99	1.09	1.88
17p12del_HNPP	0.0061	0.0016	0.037	1.97	2.25	1.23	3.00	1.38	3.50
10q11.21q11.23dup	0.0073	0.014	0.12	3.93	3.66	1.51	8.76	1.33	8.66
15q11q13del_BP3_BP4	0.0092	0.030	0.21	6.94	5.00	1.74	21.45	1.20	16.77
3q29del	0.049	0.089	0.39	5.57	5.35	1.01	21.91	0.75	26.38
16p11.2distal_dup	0.051	0.12	0.42	0.35	0.42	0.07	1.00	0.09	1.22
22q11.2del	0.063	0.22	0.54	5.03	3.58	0.90	19.98	0.42	18.55
TAR_dup	0.068	0.095	0.38	1.44	1.41	0.97	2.04	0.94	2.05
3q29dup	0.069	0.52	0.82	8.24	1.99	0.81	46.69	0.18	13.21
22q11.2distal_del	0.12	0.092	0.38	5.94	7.76	0.56	35.70	0.67	56.60
10q11.21q11.23del	0.12	0.092	0.39	2.04	2.27	0.80	4.42	0.86	5.10
NRXN1del	0.18	0.52	0.83	1.54	1.23	0.81	2.65	0.63	2.18
17q12dup	0.20	0.43	0.77	1.61	1.36	0.76	3.02	0.62	2.67
22q11.2dup	0.21	0.98	0.99	1.35	0.99	0.83	2.09	0.59	1.59
15q11.2del	0.22	0.38	0.72	1.14	1.10	0.92	1.39	0.89	1.36
2q21.1dup	0.25	0.19	0.65	0.43	0.37	0.05	1.60	0.04	1.50
15q11q13dup_BP3_BP5	0.25	0.30	0.52	3.32	2.92	0.34	15.84	0.29	14.74
7q11.23dup_distal	0.29	0.44	0.76	2.23	1.78	0.44	7.13	0.34	6.06
2q11.2dup	0.32	0.30	0.46	2.09	2.22	0.42	6.56	0.42	7.55
17q12del	0.32	0.14	0.66	2.78	5.09	0.28	14.39	0.50	28.06
15q11q13dup_BP3_BP4	0.38	0.52	0.83	0.52	0.59	0.06	1.94	0.06	2.39
15q13.3dup_CHRNA7	0.39	0.58	0.88	1.07	1.05	0.91	1.26	0.88	1.24
15q11.2dup	0.42	0.63	0.91	1.08	1.05	0.89	1.31	0.86	1.28
16p13.11del	0.44	0.63	0.91	1.34	1.21	0.61	2.57	0.53	2.42
2q21.1del	0.46	0.40	0.74	0.57	0.52	0.06	2.16	0.06	2.07
17p12dup_CMT1A	0.51	0.69	0.92	1.28	1.17	0.58	2.46	0.49	2.40
1q21.1del	0.59	0.44	0.77	0.78	0.69	0.26	1.80	0.22	1.66
22q11.2distal_dup)	0.63	0.69	0.92	0.53	0.58	0.00	4.16	0.00	4.84
WBS_dup	0.68	0.64	0.91	0.58	0.54	0.00	4.49	0.00	4.29
13q12del_CRYL1	0.69	0.64	0.92	0.90	0.89	0.53	1.44	0.50	1.45
15q24dup	0.71	0.88	0.98	1.79	1.25	0.01	14.63	0.01	10.49
15q13.3dup	0.72	0.77	0.95	1.10	1.08	0.62	1.80	0.60	1.81

2q13del_NPHP1	0.74	0.34	0.68	0.97	0.91	0.80	1.16	0.75	1.10
15q13.3del_CHRNA7	0.75	0.60	0.90	0.65	0.50	0.00	5.29	0.00	4.41
2q13del	0.79	0.32	0.67	0.84	0.54	0.17	2.53	0.11	1.70
17q11.2del_NF1	0.80	0.98	0.50	0.70	1.03	0.01	5.75	0.01	8.30
Potocki_Lupski	0.80	0.84	0.99	1.50	0.74	0.01	14.11	0.01	7.88
16p11.2dup	0.80	0.17	0.97	1.10	1.72	0.50	2.09	0.78	3.33
PWS_dup	0.80	0.77	0.95	0.71	0.68	0.01	5.52	0.01	5.23
2q13dup_NPHP1	0.82	0.76	0.92	1.02	1.03	0.83	1.24	0.83	1.26
13q12.12dup	0.82	0.61	0.95	0.93	1.19	0.47	1.65	0.59	2.13
2q11.2del	0.83	0.21	0.53	0.84	0.34	0.09	3.28	0.03	1.69
8p23.1dup	0.84	0.96	0.99	1.36	1.08	0.01	12.41	0.01	11.46
13q12.12del	0.89	0.99	0.99	0.93	1.01	0.31	2.18	0.33	2.38
TAR_del	0.89	0.83	0.97	0.93	0.88	0.25	2.40	0.24	2.36
16p12.1dup	0.97	0.74	0.95	1.01	1.12	0.51	1.81	0.55	2.04
13q12dup_CRYL1	0.98	0.76	0.96	0.97	1.62	0.01	7.86	0.01	13.51
10q23dup	0.99	0.94	1	0.98	1.11	0.01	8.93	0.01	9.68

**Supplementary Figure 6. Effect of CNVs on type 2 diabetes before and after correction for BMI, based on the above table.** It shows the changes in ORs after correction for BMI: some show increased ORs after correction for BMI. The CNVs are ordered according to the strength of the original p-value (strongest on the left).



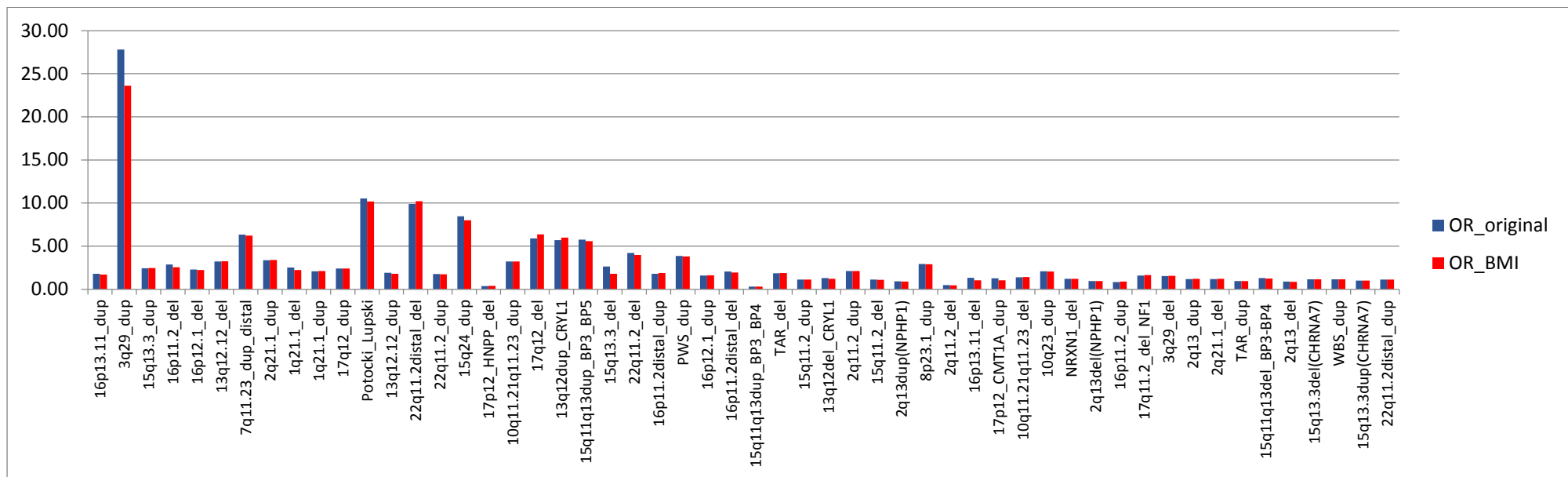
**Supplementary Table 13. Effect of all CNVs on mortality during the follow-up period, before and after correction with BMI.** The ORs remain almost unchanged. Significant results at FDR=0.1 are shown in red, with two more becoming significant after correction for BMI.

Phenotype	p-value original	p-value BMI	B-H FDR	OR original	OR BMI	OR Lower 95% original	OR Upper 95%_original	OR Lower 95%_BMI	OR Upper 95%_BMI
16p13.11_dup	0.00097	0.0031	0.050	1.79	1.70	1.21	2.32	1.28	2.43
3q29_dup	0.0013	0.0020	0.037	27.81	23.62	3.82	124.78	4.54	146.05
15q13.3_dup	0.0017	0.0015	0.041	2.42	2.46	1.45	3.90	1.43	3.85
16p11.2_del	0.0050	0.012	0.11	2.88	2.54	1.25	4.66	1.42	5.27
16p12.1_del	0.0052	0.0066	0.087	2.29	2.23	1.27	3.62	1.31	3.72
13q12.12_del	0.0092	0.0087	0.10	3.21	3.25	1.40	6.48	1.38	6.41
7q11.23_dup_distal	0.010	0.011	0.11	6.34	6.21	1.63	17.45	1.67	17.81
2q21.1_dup	0.021	0.021	0.16	3.37	3.40	1.24	7.59	1.23	7.51
1q21.1_del	0.031	0.080	0.37	2.53	2.22	0.90	4.55	1.10	5.00
1q21.1_dup	0.038	0.037	0.26	2.09	2.10	1.05	3.75	1.05	3.73
17q12_dup	0.040	0.042	0.27	2.42	2.40	1.04	4.75	1.04	4.78
Potocki_Lupski	0.048	0.050	0.29	10.52	10.17	1.00	56.33	1.02	60.19
13q12.12_dup	0.051	0.093	0.38	1.92	1.79	0.90	3.17	1.00	3.33
22q11.2distal_del	0.052	0.050	0.29	9.90	10.21	1.00	58.15	0.97	55.97
22q11.2_dup	0.053	0.062	0.34	1.77	1.73	0.97	2.84	0.99	2.90
15q24_dup	0.061	0.066	0.35	8.45	8.00	0.83	37.33	0.88	39.62
17p12_HNPP_del	0.071	0.076	0.36	0.38	0.38	0.08	1.09	0.08	1.07
10q11.21q11.23_dup	0.075	0.075	0.36	3.21	3.22	0.87	8.50	0.87	8.49
17q12_del	0.11	0.097	0.38	5.90	6.35	0.65	31.22	0.61	28.99
13q12dup_CRYL1	0.11	0.100	0.40	5.68	6.00	0.64	26.27	0.61	24.93
15q11q13dup_BP3_BP5	0.11	0.11	0.37	5.75	5.58	0.58	25.76	0.60	26.42
15q13.3_del	0.13	0.41	0.75	2.63	1.79	0.36	5.44	0.71	7.02
22q11.2_del	0.17	0.19	0.51	4.21	3.98	0.42	18.06	0.44	18.98
16p11.2distal_dup	0.18	0.16	0.49	1.80	1.87	0.76	3.81	0.73	3.67
PWS_dup	0.19	0.19	0.51	3.87	3.81	0.41	15.57	0.42	15.85
16p12.1_dup	0.22	0.20	0.52	1.59	1.62	0.75	3.04	0.73	2.98
16p11.2distal_del	0.24	0.27	0.61	2.06	1.95	0.53	5.03	0.57	5.32
15q11q13dup_BP3_BP4	0.30	0.32	0.66	0.31	0.31	0.00	2.19	0.00	2.14
TAR_del	0.31	0.30	0.65	1.84	1.88	0.52	4.80	0.51	4.71
15q11.2_dup	0.38	0.35	0.69	1.12	1.13	0.87	1.45	0.86	1.43
13q12del_CRYL1	0.39	0.53	0.83	1.29	1.21	0.63	2.09	0.70	2.18
2q11.2_dup	0.43	0.42	0.76	2.10	2.12	0.24	8.23	0.23	8.14
15q11.2_del	0.47	0.53	0.83	1.11	1.10	0.82	1.44	0.83	1.45
2q13dup(NPHP1)	0.49	0.37	0.71	0.90	0.88	0.65	1.16	0.67	1.19
8p23.1_dup	0.53	0.53	0.84	2.94	2.90	0.02	26.19	0.02	26.43
2q11.2_del	0.55	0.52	0.84	0.47	0.45	0.00	3.23	0.00	3.40
16p13.11_del	0.57	0.95	0.99	1.33	1.04	0.29	2.60	0.44	3.04
17p12_CMT1A_dup	0.63	0.95	0.91	1.28	1.03	0.29	2.60	0.42	2.94
10q11.21q11.23_del	0.63	0.62	0.99	1.40	1.41	0.29	4.15	0.29	4.11
10q23_dup	0.65	0.66	0.90	2.08	2.06	0.02	17.49	0.02	17.72
NRXN1_del	0.67	0.68	0.92	1.21	1.20	0.45	2.57	0.45	2.59
2q13del(NPHP1)	0.69	0.65	0.91	0.95	0.94	0.73	1.20	0.73	1.21
16p11.2_dup	0.71	0.81	0.96	0.82	0.88	0.24	2.20	0.23	2.06



17q11.2_del_NF1	0.77	0.75	0.95	1.59	1.64	0.01	13.01	0.01	12.66
3q29_del	0.78	0.78	0.96	1.53	1.55	0.01	12.40	0.01	12.28
2q13_dup	0.81	0.77	0.96	1.17	1.22	0.25	3.54	0.24	3.40
2q21.1_del	0.85	0.83	0.96	1.17	1.20	0.13	4.52	0.13	4.40
TAR_dup	0.86	0.88	0.98	0.95	0.95	0.50	1.64	0.50	1.63
15q11q13del_BP3-BP4	0.87	0.88	0.99	1.29	1.25	0.01	9.46	0.01	9.76
2q13_del	0.89	0.85	0.97	0.89	0.85	0.10	3.18	0.10	3.32
15q13.3del(CHRNA7)	0.92	0.93	1	1.17	1.14	0.01	9.15	0.01	9.32
WBS_dup	0.92	0.93	0.99	1.16	1.15	0.01	8.76	0.01	8.89
15q13.3dup(CHRNA7)	0.92	0.98	1	1.01	1.00	0.79	1.24	0.80	1.25
22q11.2distal_dup	0.94	0.93	1	1.11	1.13	0.01	8.75	0.01	8.61

**Supplementary Figure 7. Effect of CNVs on mortality before and after correction for BMI, based on the above table.** It shows the changes in ORs after correction for BMI: they are essentially unchanged. The CNVs are ordered according to the strength of the original p-value (strongest on the left). The first 12 CNVs were nominally significant in the original analysis.



**Supplementary Table 14. Occurrence of two CNVs in the same person.** The table lists all observed occurrences of two CNVs found in one person. CNVs that were not found in combinations with any other CNV are not shown. Over two thirds of the observations are with one of the five relatively common CNVs: 15q13.3dup(CHRNA7), 15q11.2dup/del and 2q13(NPHP1)dup/del. The observed numbers do not differ from chance expectation.

[illegible]

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
10q11.21q11.23_del	paralytic syndromes	379546	54	1962	2	0.3	6.95	0.015	7.91	1.63	23.27	159	0.30
10q11.21q11.23_del	heartfailure	376430	53	5078	3	0.7	4.03	0.028	4.53	1.21	12.15	237	0.37
10q11.21q11.23_del	stroke_derived2017	372686	52	8822	4	1.2	3.09	0.040	3.31	1.07	7.96	291	0.43
10q11.21q11.23_del	anaemia	362362	50	19146	6	2.6	2.14	0.089	2.20	0.87	4.65	483	0.58
10q11.21q11.23_del	heart_other	369683	52	11825	4	1.7	2.30	0.091	2.60	0.84	6.25	493	0.58
10q11.21q11.23_del	aneurism	379541	55	1967	1	0.3	3.46	0.12	4.99	0.56	19.05	615	0.63
10q11.21q11.23_del	diabetes_other	360768	50	20740	6	2.9	1.97	0.12	2.04	0.80	4.42	617	0.63
10q11.21q11.23_del	irritable_bowel_syndrome	368501	52	13007	4	1.8	2.10	0.14	2.27	0.74	5.37	661	0.65
10q11.21q11.23_del	arrhythmia	357344	50	24164	6	3.4	1.69	0.16	1.90	0.75	4.06	730	0.70
10q11.21q11.23_del	psoriasis	375486	54	6022	2	0.9	2.26	0.18	2.74	0.57	8.01	772	0.72
10q11.21q11.23_del	gout	374416	54	7092	2	1.0	1.92	0.20	2.62	0.53	8.03	850	0.74
10q11.21q11.23_del	glaucoma	374978	54	6530	2	0.9	2.09	0.20	2.58	0.53	7.67	856	0.74
10q11.21q11.23_del	nasal	378315	55	3193	1	0.5	2.13	0.21	3.45	0.39	12.74	880	0.75
10q11.21q11.23_del	low_WBC_count	378542	55	2966	1	0.4	2.30	0.22	3.34	0.38	12.30	905	0.77
10q11.21q11.23_del	ocular	359843	51	21665	5	3.1	1.57	0.25	1.74	0.64	3.87	1011	0.79
10q11.21q11.23_del	ischaemic_heart_disease_not_MI	365181	52	16327	4	2.3	1.67	0.26	1.85	0.59	4.48	1027	0.79
10q11.21q11.23_del	neuropathies	365601	52	15907	4	2.3	1.71	0.28	1.78	0.58	4.20	1095	0.80
10q11.21q11.23_del	ovarian cysts	374118	54	7390	2	1.1	1.84	0.31	2.11	0.43	6.36	1176	0.81
10q11.21q11.23_del	osteoporosis	369912	53	11596	3	1.7	1.76	0.33	1.83	0.49	4.90	1250	0.82
10q11.21q11.23_del	heart_valve	373130	54	8378	2	1.2	1.63	0.33	2.01	0.41	5.92	1257	0.82
10q11.21q11.23_del	gastric_reflux	347206	49	34302	7	4.8	1.39	0.34	1.48	0.63	3.02	1298	0.83
10q11.21q11.23_del	cerebrovascular_disease_not_stroke	377177	55	4331	1	0.6	1.57	0.37	2.32	0.26	8.68	1375	0.85
10q11.21q11.23_del	venous_thromboembolic_disease	367533	53	13975	3	2.0	1.46	0.44	1.58	0.43	4.07	1576	0.87
10q11.21q11.23_del	high_cholesterol	314916	44	66592	12	9.3	1.23	0.45	1.30	0.64	2.45	1605	0.87
10q11.21q11.23_del	connective tissue disorder	372154	54	9354	2	1.4	1.46	0.48	1.64	0.34	4.83	1706	0.88
10q11.21q11.23_del	congenital	376480	55	5028	1	0.7	1.36	0.49	1.87	0.21	6.86	1738	0.89
10q11.21q11.23_del	gastrointestinal ulcer	371154	54	10354	2	1.5	1.32	0.53	1.55	0.32	4.58	1839	0.90
10q11.21q11.23_del	asthma	332175	47	49333	9	7.0	1.24	0.54	1.25	0.59	2.41	1853	0.91
10q11.21q11.23_del	cataract	359656	54	21852	2	3.3	0.62	0.55	0.69	0.14	2.07	1884	0.91
10q11.21q11.23_del	biliary	360730	52	20778	4	3.0	1.31	0.56	1.35	0.44	3.21	1915	0.91
10q11.21q11.23_del	sciatica	338392	51	43116	5	6.5	0.79	0.59	0.79	0.29	1.75	1990	0.93
10q11.21q11.23_del	ear	367288	55	14220	1	2.1	0.48	0.60	0.66	0.08	2.45	2007	0.93
10q11.21q11.23_del	osteoarthritis	312791	44	68717	12	9.7	1.19	0.60	1.19	0.60	2.22	2016	0.94
10q11.21q11.23_del	prostate hyperplasia	367059	54	14449	2	2.1	0.94	0.61	1.44	0.28	4.75	2050	0.94
10q11.21q11.23_del	thyroid	354365	53	27143	3	4.1	0.75	0.61	0.76	0.21	1.99	2051	0.94
10q11.21q11.23_del	died_to_2016	370537	54	10971	2	1.6	1.24	0.63	1.40	0.29	4.11	2081	0.94
10q11.21q11.23_del	atherosclerotic_vascular_disease	375758	55	5750	1	0.8	1.18	0.63	1.53	0.17	5.76	2097	0.94
10q11.21q11.23_del	varicose_veins	369654	54	11854	2	1.7	1.15	0.66	1.35	0.28	3.94	2179	0.95
10q11.21q11.23_del	digestive	299653	45	81855	11	12.3	0.92	0.67	0.87	0.43	1.61	2203	0.95
10q11.21q11.23_del	COPD	367277	53	14231	3	2.1	1.44	0.67	1.29	0.34	3.45	2210	0.95
10q11.21q11.23_del	ureter_bladder	332973	48	48535	8	7.0	1.12	0.69	1.16	0.52	2.30	2273	0.95
10q11.21q11.23_del	diverticular_disease_intestine	354372	53	27136	3	4.1	0.75	0.70	0.81	0.22	2.11	2287	0.96
10q11.21q11.23_del	any_Ca	331806	50	49702	6	7.5	0.82	0.72	0.86	0.34	1.84	2364	0.96
10q11.21q11.23_del	inflammatory_bowel_disease	362916	54	18592	2	2.8	0.73	0.74	0.81	0.17	2.38	2415	0.97
10q11.21q11.23_del	hernia	333614	50	47894	6	7.2	0.85	0.75	0.88	0.35	1.87	2429	0.97
10q11.21q11.23_del	renal	374218	55	7290	1	1.1	0.93	0.77	1.29	0.15	4.74	2490	0.97
10q11.21q11.23_del	allergy	322839	47	58669	9	8.5	1.05	0.78	1.10	0.52	2.12	2533	0.97
10q11.21q11.23_del	migraine	367828	54	13680	2	2.0	1.00	0.80	1.19	0.24	3.49	2584	0.97
10q11.21q11.23_del	hypertension	260858	37	120650	19	17.1	1.07	0.81	1.07	0.59	1.88	2614	0.97
10q11.21q11.23_del	obesity	371670	55	9838	1	1.5	0.69	0.84	0.85	0.10	3.13	2698	0.97
10q11.21q11.23_del	uterine_problem	339214	50	42294	6	6.2	0.97	0.85	0.92	0.36	2.04	2731	0.97
10q11.21q11.23_del	MI_derived2017	367646	54	13862	2	2.0	0.98	0.85	1.14	0.23	3.49	2732	0.97
10q11.21q11.23_del	endocrine_other	379148	56	2360	0	0.3	0.00	0.86	1.29	0.01	8.95	2777	0.98
10q11.21q11.23_del	renal_failure	373587	55	7921	1	1.2	0.86	0.89	1.13	0.13	4.23	2829	0.98
10q11.21q11.23_del	coagulation_defects	378967	56	2541	0	0.4	0.00	0.89	1.22	0.01	8.49	2846	0.98
10q11.21q11.23_del	diabetes_insulin_dependent	378914	56	2594	0	0.4	0.00	0.94	1.11	0.01	7.73	2994	0.99
10q11.21q11.23_del	respiratory	360755	53	20753	3	3.0	0.98	0.95	1.04	0.28	2.68	3009	0.99
10q11.21q11.23_del	hepatic	372754	55	8754	1	1.3	0.78	0.97	1.03	0.12	3.79	3074	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
10q11.21q11.23_dup	diabetes_other	360752	34	20740	6	2.0	2.76	0.0073	3.93	1.51	8.76	101	0.23
10q11.21q11.23_dup	varicose_veins	369636	36	11856	4	1.2	3.22	0.013	4.52	1.45	10.92	140	0.28
10q11.21q11.23_dup	biliary	360713	35	20779	5	2.0	2.30	0.020	3.48	1.25	8.02	184	0.35
10q11.21q11.23_dup	osteoporosis	369896	37	11596	3	1.2	2.47	0.027	4.77	1.23	13.68	231	0.37
10q11.21q11.23_dup	asthma	332167	39	49325	1	5.8	0.19	0.039	0.25	0.03	0.94	284	0.43
10q11.21q11.23_dup	neuropathies	365585	36	15907	4	1.6	2.40	0.043	3.25	1.04	7.88	304	0.45
10q11.21q11.23_dup	atherosclerotic_vascular_disease	375741	38	5751	2	0.6	3.32	0.046	5.14	1.04	15.66	314	0.45
10q11.21q11.23_dup	obesity	371652	37	9840	3	1.0	2.91	0.046	3.79	1.03	10.01	316	0.46
10q11.21q11.23_dup	hypertension	260845	24	120647	16	11.1	1.26	0.054	1.94	0.99	3.71	348	0.49
10q11.21q11.23_dup	died_to_2016	370520	37	10972	3	1.1	2.61	0.075	3.21	0.87	8.49	426	0.55
10q11.21q11.23_dup	MI_derived2017	367629	37	13863	3	1.4	2.06	0.090	3.09	0.81	8.49	489	0.58
10q11.21q11.23_dup	heart_valve	373114	38	8378	2	0.9	2.28	0.10	3.57	0.73	10.75	534	0.60
10q11.21q11.23_dup	ocular	359828	36	21664	4	2.2	1.76	0.11	2.48	0.79	6.02	545	0.62
10q11.21q11.23_dup	migraine	367811	37	13681	3	1.4	2.09	0.12	2.71	0.73	7.20	603	0.63
10q11.21q11.23_dup	nasal	378299	39	3193	1	0.3	2.99	0.13	4.85	0.55	18.16	628	0.64
10q11.21q11.23_dup	respiratory	360738	36	20754	4	2.1	1.84	0.13	2.34	0.75	5.69	634	0.64
10q11.21q11.23_dup	ureter_bladder	332958	33	48534	7	4.8	1.38	0.18	1.78	0.74	3.76	790	0.73
10q11.21q11.23_dup	inflammatory_bowel_disease	362902	40	18590	0	2.0	0.00	0.23	0.26	0.00	1.82	925	0.77
10q11.21q11.23_dup	high_cholesterol	314904	32	66588	8	6.8	1.15	0.23	1.66	0.70	3.54	938	0.78
10q11.21q11.23_dup	gastric_reflux	347192	35	34300	5	3.5	1.39	0.24	1.79	0.65	4.08	959	0.78
10q11.21q11.23_dup	prostate_hyperplasia	367043	38	14449	2	1.5	1.32	0.25	2.42	0.47	8.06	1007	0.78
10q11.21q11.23_dup	glaucoma	374963	39	6529	1	0.7	1.46	0.27	2.94	0.33	11.24	1057	0.80
10q11.21q11.23_dup	ovarian_cysts	374103	39	7389	1	0.8	1.29	0.34	2.54	0.28	10.23	1272	0.83
10q11.21q11.23_dup	ear	367273	40	14219	0	1.5	0.00	0.38	0.35	0.00	2.49	1413	0.85
10q11.21q11.23_dup	renal_failure	373571	39	7921	1	0.8	1.20	0.41	2.17	0.24	8.32	1494	0.86
10q11.21q11.23_dup	allergy	322828	36	58664	4	6.5	0.65	0.44	0.69	0.22	1.66	1578	0.87
10q11.21q11.23_dup	gout	374401	39	7091	1	0.7	1.35	0.45	2.02	0.22	7.78	1618	0.87
10q11.21q11.23_dup	COPD	367264	40	14228	0	1.5	0.00	0.46	0.40	0.00	2.92	1637	0.88
10q11.21q11.23_dup	aneurism	379526	40	1966	0	0.2	0.00	0.49	3.20	0.03	23.03	1740	0.89
10q11.21q11.23_dup	cataract	359640	38	21852	2	2.3	0.87	0.51	1.60	0.32	5.04	1798	0.90
10q11.21q11.23_dup	heart_other	369671	40	11821	0	1.3	0.00	0.53	0.46	0.00	3.25	1843	0.90
10q11.21q11.23_dup	hernia	333601	37	47891	3	5.3	0.60	0.54	0.72	0.19	1.90	1857	0.91
10q11.21q11.23_dup	osteoarthritis	312783	36	68709	4	7.9	0.56	0.54	0.74	0.23	1.84	1870	0.91
10q11.21q11.23_dup	digestive	299642	34	81850	6	9.3	0.70	0.56	0.78	0.30	1.70	1900	0.92
10q11.21q11.23_dup	uterine_problem	339200	36	42292	4	4.5	0.90	0.57	1.38	0.42	3.80	1934	0.92
10q11.21q11.23_dup	hepatic	372738	39	8754	1	0.9	1.09	0.58	1.64	0.18	6.15	1953	0.93
10q11.21q11.23_dup	paralytic_syndromes	379532	40	1960	0	0.2	0.00	0.60	2.36	0.02	16.64	2006	0.93
10q11.21q11.23_dup	diverticular_disease_intestine	354358	39	27134	1	3.0	0.35	0.60	0.66	0.07	2.53	2019	0.94
10q11.21q11.23_dup	endocrine_other	379132	40	2360	0	0.2	0.00	0.61	2.27	0.02	15.99	2045	0.94
10q11.21q11.23_dup	gastrointestinal_ulcer	371139	39	10353	1	1.1	0.92	0.64	1.52	0.17	5.75	2121	0.94
10q11.21q11.23_dup	sciatica	338378	37	43114	3	4.7	0.66	0.65	0.78	0.21	2.06	2164	0.95
10q11.21q11.23_dup	coagulation_defects	378951	40	2541	0	0.3	0.00	0.68	1.91	0.02	13.38	2242	0.95
10q11.21q11.23_dup	low_WBC_count	378527	40	2965	0	0.3	0.00	0.71	1.77	0.01	12.40	2320	0.96
10q11.21q11.23_dup	connective_tissue_disorder	372140	40	9352	0	1.0	0.00	0.72	0.62	0.00	4.41	2336	0.96
10q11.21q11.23_dup	stroke_derived2017	372674	40	8818	0	0.9	0.00	0.72	0.62	0.00	4.45	2341	0.96
10q11.21q11.23_dup	diabetes_insulin_dependent	378898	40	2594	0	0.3	0.00	0.72	1.74	0.01	12.31	2346	0.96
10q11.21q11.23_dup	renal	374203	40	7289	0	0.8	0.00	0.77	0.68	0.01	4.78	2508	0.97
10q11.21q11.23_dup	arrhythmia	357332	38	24160	2	2.6	0.79	0.80	1.19	0.24	3.60	2574	0.97
10q11.21q11.23_dup	anaemia	362351	39	19141	1	2.1	0.50	0.80	0.82	0.09	3.06	2593	0.97
10q11.21q11.23_dup	cerebrovascular_disease_not_stroke	377162	40	4330	0	0.5	0.00	0.82	1.42	0.01	10.15	2632	0.97
10q11.21q11.23_dup	psoriasis	375472	40	6020	0	0.6	0.00	0.82	0.73	0.01	5.14	2634	0.97
10q11.21q11.23_dup	irritable_bowel_syndrome	368488	39	13004	1	1.4	0.73	0.84	1.19	0.13	4.48	2697	0.97
10q11.21q11.23_dup	venous_thromboembolic_disease	367519	39	13973	1	1.5	0.68	0.86	1.16	0.13	4.37	2765	0.97
10q11.21q11.23_dup	thyroid	354350	38	27142	2	2.9	0.70	0.89	1.10	0.22	3.39	2842	0.98
10q11.21q11.23_dup	any_Ca	331792	36	49700	4	5.4	0.77	0.89	1.07	0.34	2.61	2857	0.98
10q11.21q11.23_dup	ischaemic_heart_disease_not_MI	365168	39	16324	1	1.7	0.58	0.93	1.08	0.12	4.17	2950	0.98
10q11.21q11.23_dup	heartfailure	376417	40	5075	0	0.5	0.00	0.94	1.12	0.01	8.14	2973	0.99
10q11.21q11.23_dup	congenital	376465	40	5027	0	0.5	0.00	0.95	0.92	0.01	6.43	3020	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
10q23_dup	cerebrovascular_disease_not_stroke	377128	6	4331	1	0.1	12.59	0.013	24.47	2.35	137.34	143	0.28
10q23_dup	congenital	376431	6	5028	1	0.1	10.84	0.021	16.58	1.72	79.30	190	0.35
10q23_dup	atherosclerotic_vascular_disease	375709	6	5750	1	0.1	9.48	0.024	16.39	1.61	88.70	209	0.35
10q23_dup	asthma	332132	4	49327	3	0.6	3.31	0.032	5.27	1.18	21.69	252	0.39
10q23_dup	heart_valve	373082	6	8377	1	0.1	6.51	0.039	11.66	1.17	60.16	286	0.43
10q23_dup	hepatic	372705	6	8754	1	0.1	6.23	0.051	9.60	0.98	47.19	333	0.48
10q23_dup	gastrointestinal_ulcer	371106	6	10353	1	0.2	5.26	0.053	9.61	0.96	49.66	343	0.49
10q23_dup	gastric_reflux	347162	5	34297	2	0.5	3.18	0.055	5.44	0.96	23.29	353	0.49
10q23_dup	venous_thromboembolic_disease	367486	6	13973	1	0.2	3.90	0.090	6.69	0.68	33.35	491	0.58
10q23_dup	ear	367239	6	14220	1	0.2	3.83	0.10	6.19	0.64	30.31	527	0.60
10q23_dup	neuropathies	365555	6	15904	1	0.3	3.43	0.11	5.83	0.60	28.55	554	0.62
10q23_dup	digestive	299612	4	81847	3	1.1	2.00	0.11	3.38	0.73	14.49	564	0.63
10q23_dup	cataract	359608	6	21851	1	0.4	2.49	0.13	5.70	0.53	34.62	638	0.64
10q23_dup	anaemia	362318	6	19141	1	0.3	2.85	0.14	4.97	0.51	24.37	666	0.66
10q23_dup	ureter_bladder	332930	5	48529	2	0.7	2.25	0.14	3.55	0.62	15.48	668	0.66
10q23_dup	prostate_hyperplasia	367011	6	14448	1	0.2	3.77	0.17	5.07	0.44	38.58	744	0.71
10q23_dup	diverticular_disease_intestine	354325	6	27134	1	0.5	2.01	0.19	4.10	0.40	21.91	816	0.74
10q23_dup	endocrine_other	379099	7	2360	0	0.0	0.00	0.21	12.62	0.10	105.94	871	0.75
10q23_dup	aneurism	379493	7	1966	0	0.0	0.00	0.22	11.88	0.09	115.77	902	0.76
10q23_dup	arrhythmia	357300	6	24159	1	0.4	2.26	0.22	3.70	0.36	19.69	914	0.77
10q23_dup	osteoarthritis	312752	5	68707	2	1.1	1.59	0.23	2.84	0.48	13.22	919	0.77
10q23_dup	paralytic_syndromes	379499	7	1960	0	0.0	0.00	0.23	10.91	0.08	93.48	930	0.77
10q23_dup	low_WBC_count	378494	7	2965	0	0.1	0.00	0.24	10.18	0.08	84.17	958	0.78
10q23_dup	coagulation_defects	378918	7	2541	0	0.0	0.00	0.25	9.63	0.07	80.51	992	0.78
10q23_dup	allergy	322797	5	58662	2	0.9	1.86	0.27	2.46	0.44	10.24	1061	0.80
10q23_dup	diabetes_insulin_dependent	378865	7	2594	0	0.0	0.00	0.28	7.79	0.06	66.90	1108	0.80
10q23_dup	nasal	378267	7	3192	0	0.1	0.00	0.28	7.74	0.06	64.38	1110	0.80
10q23_dup	high_cholesterol	314877	5	66582	2	1.1	1.64	0.31	2.47	0.38	12.68	1206	0.81
10q23_dup	hypertension	260825	4	120634	3	1.9	1.36	0.38	2.01	0.40	9.43	1411	0.85
10q23_dup	osteoporosis	369866	7	11593	0	0.2	0.00	0.41	4.47	0.03	42.18	1487	0.86
10q23_dup	psoriasis	375439	7	6020	0	0.1	0.00	0.43					

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
13q12.12_del	sciatica	338406	65	43130	19	8.3	2.00	0.0027	2.32	1.36	3.78	62	0.14
13q12.12_del	died to 2016	370560	77	10976	7	2.3	2.90	0.0092	3.21	1.38	6.41	117	0.25
13q12.12_del	hypertension	260867	46	120669	38	21.3	1.43	0.014	1.77	1.12	2.77	152	0.29
13q12.12_del	low WBC count	378568	81	2968	3	0.6	4.59	0.014	5.40	1.50	13.62	153	0.29
13q12.12_del	high cholesterol	314933	61	66603	23	12.9	1.57	0.031	1.78	1.06	2.92	246	0.39
13q12.12_del	uterine problem	339233	69	42303	15	8.6	1.61	0.044	1.93	1.02	3.50	308	0.45
13q12.12_del	any Ca	331823	67	49713	17	10.0	1.55	0.079	1.66	0.94	2.77	450	0.55
13q12.12_del	gastric reflux	347229	72	34307	12	7.1	1.59	0.11	1.70	0.89	3.00	540	0.62
13q12.12_del	gastrointestinal ulcer	371184	84	10352	0	2.3	0.00	0.11	0.19	0.00	1.33	570	0.63
13q12.12_del	heart valve	373156	80	8380	4	1.8	2.17	0.11	2.38	0.78	5.53	572	0.63
13q12.12_del	respiratory	360778	76	20758	8	4.4	1.75	0.12	1.86	0.84	3.59	579	0.63
13q12.12_del	connective tissue disorder	372180	80	9356	4	2.0	1.94	0.15	2.17	0.71	5.05	703	0.69
13q12.12_del	glaucoma	375005	81	6531	3	1.4	2.09	0.17	2.34	0.64	5.96	751	0.72
13q12.12_del	hernia	333641	77	47895	7	11.1	0.66	0.20	0.63	0.27	1.25	837	0.74
13q12.12_del	paralytic syndromes	379575	83	1961	1	0.4	2.32	0.21	3.42	0.39	12.46	886	0.76
13q12.12_del	ischaemic heart disease not MI	365207	78	16329	6	3.5	1.67	0.23	1.71	0.68	3.60	931	0.77
13q12.12_del	venous thromboembolic disease	367559	79	13977	5	3.0	1.63	0.24	1.76	0.65	3.82	963	0.78
13q12.12_del	COPD	367303	79	14233	5	3.1	1.60	0.27	1.70	0.62	3.78	1065	0.80
13q12.12_del	renal	374247	84	7289	0	1.6	0.00	0.28	0.29	0.00	2.02	1099	0.80
13q12.12_del	MI derived2017	367671	79	13865	5	3.0	1.64	0.28	1.69	0.61	3.77	1100	0.80
13q12.12_del	renal failure	373613	81	7923	3	1.7	1.72	0.29	1.90	0.52	4.85	1119	0.80
13q12.12_del	coagulation defects	378994	83	2542	1	0.6	1.79	0.30	2.71	0.31	9.84	1148	0.81
13q12.12_del	osteoporosis	369939	80	11597	4	2.5	1.57	0.30	1.74	0.56	4.15	1165	0.81
13q12.12_del	osteoarthritis	312819	72	68717	12	15.8	0.79	0.31	0.74	0.38	1.31	1195	0.81
13q12.12_del	migraine	367857	83	13679	1	3.1	0.33	0.32	0.48	0.06	1.77	1233	0.82
13q12.12_del	hepatic	372780	81	8756	3	1.9	1.56	0.33	1.79	0.49	4.51	1256	0.82
13q12.12_del	thyroid	354388	76	27148	8	5.8	1.34	0.34	1.44	0.65	2.82	1296	0.83
13q12.12_del	ureter bladder	333001	76	48535	8	11.1	0.75	0.36	0.72	0.33	1.40	1330	0.84
13q12.12_del	atherosclerotic vascular disease	375785	82	5751	2	1.3	1.58	0.37	1.89	0.39	5.51	1364	0.85
13q12.12_del	heart other	369714	83	11822	1	2.7	0.38	0.37	0.52	0.06	1.89	1382	0.85

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
13q12.12_dup	died to 2016	370684	201	10980	11	6.0	1.80	0.051	1.92	1.00	3.33	334	0.48
13q12.12_dup	high_cholesterol	315058	186	66606	26	39.3	0.70	0.065	0.68	0.44	1.02	379	0.54
13q12.12_dup	cataract	359797	195	21867	17	11.9	1.40	0.084	1.60	0.94	2.58	466	0.56
13q12.12_dup	thyroid	354503	191	27161	21	14.6	1.39	0.096	1.50	0.93	2.32	512	0.59
13q12.12_dup	COPD	367424	200	14240	12	7.8	1.52	0.10	1.68	0.89	2.89	533	0.60
13q12.12_dup	osteoarthritis	312930	183	68734	29	40.2	0.76	0.11	0.73	0.48	1.07	563	0.63
13q12.12_dup	renal_failure	373743	211	7921	1	4.5	0.23	0.12	0.35	0.04	1.25	610	0.63
13q12.12_dup	obesity	371825	210	9839	2	5.6	0.37	0.14	0.44	0.09	1.24	659	0.65
13q12.12_dup	hypertension	260977	156	120687	56	72.1	0.84	0.15	0.80	0.58	1.09	695	0.68
13q12.12_dup	migraine	367982	208	13682	4	7.7	0.53	0.18	0.56	0.19	1.26	775	0.72
13q12.12_dup	ocular	359988	196	21676	16	11.8	1.33	0.18	1.44	0.83	2.31	779	0.72
13q12.12_dup	heart_valve	373281	205	8383	7	4.6	1.50	0.19	1.71	0.75	3.30	798	0.73
13q12.12_dup	osteoporosis	370068	209	11596	3	6.5	0.47	0.20	0.54	0.15	1.34	858	0.74
13q12.12_dup	inflammatory_bowel_disease	363060	198	18604	14	10.1	1.36	0.21	1.43	0.80	2.35	889	0.76
13q12.12_dup	MI_derived2017	367800	208	13864	4	7.8	0.52	0.24	0.60	0.20	1.36	976	0.78
13q12.12_dup	respiratory	360899	197	20765	15	11.3	1.30	0.24	1.38	0.79	2.25	978	0.78
13q12.12_dup	diverticular_disease_intestine	354513	194	27151	18	14.9	1.19	0.30	1.30	0.78	2.06	1153	0.81
13q12.12_dup	glaucoma	375131	207	6533	5	3.6	1.38	0.30	1.62	0.61	3.44	1174	0.81
13q12.12_dup	digestive	299781	173	81883	39	47.3	0.86	0.31	0.84	0.58	1.17	1203	0.82
13q12.12_dup	any_Ca	331937	181	49727	31	27.1	1.12	0.32	1.22	0.82	1.77	1218	0.82
13q12.12_dup	diabetes_insulin_dependent	379070	212	2594	0	1.5	0.00	0.35	0.34	0.00	2.31	1325	0.84
13q12.12_dup	allergy	322976	184	58688	28	33.4	0.86	0.37	0.84	0.55	1.22	1376	0.85
13q12.12_dup	sciatica	338525	184	43139	28	23.4	1.17	0.39	1.19	0.79	1.74	1437	0.85
13q12.12_dup	heart_other	369835	204	11829	8	6.5	1.22	0.39	1.37	0.63	2.57	1459	0.85
13q12.12_dup	endocrine_other	379304	212	2360	0	1.3	0.00	0.42	0.38	0.00	2.58	1504	0.87
13q12.12_dup	coagulation_defects	379121	210	2543	2	1.4	1.42	0.42	1.76	0.37	4.99	1505	0.87
13q12.12_dup	irritable_bowel_syndrome	368652	203	13012	9	7.2	1.25	0.46	1.29	0.63	2.34	1643	0.88
13q12.12_dup	atherosclerotic_vascular_disease	375911	208	5753	4	3.2	1.25	0.46	1.45	0.48	3.29	1650	0.88
13q12.12_dup	arrhythmia	357491	197	24173	15	13.3	1.12	0.47	1.22	0.69	1.99	1681	0.88
13q12.12_dup	hepatic	372905	206	8759	6	4.8	1.23	0.49	1.33	0.55	2.68	1742	0.89</

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
13q12del_CRYL1	phenotypic syndromes	379850	358	1965	5	1.9	2.68	0.023	3.18	1.20	6.71	208	0.35
13q12del_CRYL1	migraine	368131	357	13684	6	13.3	0.46	0.030	0.47	0.19	0.94	242	0.39
13q12del_CRYL1	ovarian cysts	374425	361	7390	2	7.1	0.28	0.038	0.34	0.07	0.95	276	0.43
13q12del_CRYL1	respiratory	361038	336	20777	27	19.3	1.37	0.040	1.55	1.02	2.24	287	0.43
13q12del_CRYL1	arrhythmia	357627	333	24188	30	22.5	1.30	0.049	1.49	1.00	2.13	324	0.47
13q12del_CRYL1	nasal	378623	363	3192	0	3.1	0.00	0.072	0.17	0.00	1.12	416	0.54
13q12del_CRYL1	digestive	299886	278	81929	85	75.9	1.09	0.11	1.22	0.95	1.55	568	0.63
13q12del_CRYL1	psoriasis	375786	354	6029	9	5.7	1.57	0.12	1.75	0.85	3.14	593	0.63
13q12del_CRYL1	low WBC count	378845	358	2970	5	2.8	1.77	0.14	2.03	0.76	4.26	669	0.66
13q12del_CRYL1	prostate hyperplasia	367360	355	14455	8	14.0	0.58	0.18	0.64	0.29	1.21	770	0.72
13q12del_CRYL1	heart valve	373435	359	8380	4	8.1	0.50	0.24	0.60	0.20	1.35	979	0.78
13q12del_CRYL1	ocular	360131	339	21684	24	20.4	1.16	0.27	1.27	0.82	1.88	1067	0.80
13q12del_CRYL1	inflammatory bowel disease	363204	342	18611	21	17.5	1.19	0.27	1.29	0.81	1.94	1072	0.80
13q12del_CRYL1	ischaemic heart disease_not_MI	365474	345	16341	18	15.4	1.16	0.32	1.29	0.77	2.04	1226	0.81
13q12del_CRYL1	high cholesterol	315184	312	66631	51	66.0	0.80	0.33	0.86	0.63	1.16	1251	0.82
13q12del_CRYL1	hernia	333890	326	47925	37	46.8	0.81	0.36	0.85	0.60	1.19	1335	0.84
13q12del_CRYL1	heartfailure	376738	361	5077	2	4.9	0.41	0.37	0.59	0.12	1.67	1359	0.85
13q12del_CRYL1	gastric reflux	347485	328	34330	35	32.4	1.07	0.37	1.18	0.82	1.64	1381	0.85
13q12del_CRYL1	died to 2016	370834	351	10981	12	10.4	1.15	0.39	1.29	0.70	2.18	1450	0.85
13q12del_CRYL1	any_Ca	332070	314	49745	49	47.0	1.04	0.40	1.14	0.83	1.53	1473	0.85
13q12del_CRYL1	anaemia	362655	343	19160	20	18.1	1.10	0.44	1.20	0.74	1.82	1579	0.87
13q12del_CRYL1	gastrointestinal ulcer	371457	357	10358	6	10.0	0.61	0.44	0.75	0.31	1.50	1593	0.87
13q12del_CRYL1	varicose veins	369955	355	11860	8	11.4	0.71	0.48	0.79	0.37	1.46	1702	0.88
13q12del_CRYL1	glaucoma	375280	356	6535	7	6.2	1.13	0.49	1.31	0.57	2.51	1729	0.89
13q12del_CRYL1	renal failure	373887	355	7928	8	7.5	1.06	0.51	1.27	0.59	2.36	1782	0.89
13q12del_CRYL1	cerebrovascular disease_not_stroke	377483	361	4332	2	4.1	0.49	0.51	0.68	0.14	1.91	1791	0.89
13q12del_CRYL1	neuropathies	365899	350	15916	13	15.2	0.86	0.52	0.84	0.45	1.41	1824	0.90
13q12del_CRYL1	coagulation defects	379273	362	2542	1	2.4	0.41	0.56	0.64	0.07	2.29	1913	0.91
13q12del_CRYL1	obesity	371971	356	9844	7	9.4	0.75	0.60	0.83	0.36	1.59	2003	0.93
13q12del_CRYL1	thyroid	354649	337	27166	26	25.8	1.01	0.68	1.09	0.71			



CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
13q12dup_CRYL1	inflammatory_bowel_disease	362869	7	18593	3	0.4	6.16	0.0058	8.37	2.02	28.32	86	0.21
13q12dup_CRYL1	arrhythmia	357301	7	24161	3	0.5	4.74	0.0071	8.13	1.91	28.42	98	0.23
13q12dup_CRYL1	heart_other	369639	8	11823	2	0.3	6.45	0.0091	11.24	2.05	42.70	114	0.25
13q12dup_CRYL1	low_WBC_count	378496	9	2966	1	0.1	12.87	0.016	19.00	2.03	82.75	163	0.31
13q12dup_CRYL1	migraine	367782	8	13680	2	0.3	5.58	0.020	7.98	1.49	29.66	178	0.35
13q12dup_CRYL1	heart_valve	373085	9	8377	1	0.2	4.55	0.073	7.40	0.78	32.91	418	0.55
13q12dup_CRYL1	died_to_2016	370492	9	10970	3	0.3	3.48	0.11	5.68	0.61	24.93	548	0.62
13q12dup_CRYL1	prostate_hyperplasia	367014	9	14448	1	0.4	2.64	0.13	6.78	0.52	76.51	632	0.64
13q12dup_CRYL1	osteoporosis	369868	9	11594	1	0.3	3.29	0.17	4.30	0.45	20.13	747	0.71
13q12dup_CRYL1	digestive	299614	6	81848	4	1.6	1.86	0.17	2.41	0.66	8.03	749	0.71
13q12dup_CRYL1	ischaemic_heart_disease_not_MI	365138	9	16324	1	0.4	2.34	0.18	4.21	0.44	19.50	764	0.72
13q12dup_CRYL1	gastric_reflux	347165	8	34297	2	0.8	2.22	0.18	2.92	0.55	10.63	782	0.73
13q12dup_CRYL1	aneurism	379496	10	1966	0	0.1	0.00	0.23	10.76	0.08	93.40	936	0.78
13q12dup_CRYL1	paralytic_syndromes	379502	10	1960	0	0.1	0.00	0.24	10.15	0.08	79.83	954	0.78
13q12dup_CRYL1	diabetes_insulin_dependent	378868	10	2594	0	0.1	0.00	0.28	7.70	0.06	60.93	1109	0.80
13q12dup_CRYL1	coagulation_defects	378921	10	2541	0	0.1	0.00	0.30	7.17	0.06	55.75	1144	0.81
13q12dup_CRYL1	respiratory	360711	9	20751	1	0.5	1.84	0.30	2.82	0.30	12.36	1154	0.81
13q12dup_CRYL1	ocular	359801	9	21661	1	0.5	1.76	0.30	2.82	0.30	12.43	1162	0.81
13q12dup_CRYL1	endocrine_other	379102	10	2360	0	0.1	0.00	0.30	6.96	0.05	54.46	1166	0.81
13q12dup_CRYL1	nasal	378270	10	3192	0	0.1	0.00	0.32	6.47	0.05	50.89	1216	0.82
13q12dup_CRYL1	any_Ca	331764	8	49698	2	1.2	1.54	0.39	1.96	0.36	7.33	1434	0.85
13q12dup_CRYL1	cerebrovascular_disease_not_stroke	377132	10	4330	0	0.1	0.00	0.40	4.56	0.04	36.95	1466	0.85
13q12dup_CRYL1	heartfailure	376387	10	5075	0	0.1	0.00	0.41	4.34	0.03	36.56	1498	0.86
13q12dup_CRYL1	sciatica	338351	10	43111	0	1.3	0.00	0.42	0.37	0.00	2.87	1524	0.86
13q12dup_CRYL1	hypertension	260827	6	120635	4	2.8	1.26	0.42	1.68	0.45	5.76	1529	0.87
13q12dup_CRYL1	asthma	332136	8	49326	2	1.2	1.55	0.44	1.80	0.34	6.59	1586	0.87
13q12dup_CRYL1	diverticular_disease_intestine	354328	9	27134	1	0.7	1.41	0.47	2.02	0.21	9.08	1661	0.88
13q12dup_CRYL1	congenital	376435	10	5027	0	0.1	0.00	0.48	3.38	0.03	26.29	1700	0.88
13q12dup_CRYL1	gout	374372	10	7090	0	0.2	0.00	0.48	3.42	0.03	32.83	1717	0.88
13q12dup_CRYL1	atherosclerotic_vascular_disease	375713	10	5749	0	0.2	0.00	0.49	3.28	0.03	26.05	1727	0.89
13q12dup_CRYL1	high_cholesterol	314880	8	66582	2	1.7	1.15	0.52	1.67	0.30	6.47	1802	0.90
13q12dup_CRYL1	glaucoma	374934	10	6528	0	0.2	0.00	0.52	3.03	0.02	24.90	1803	0.90
13q12dup_CRYL1	psoriasis	375442	10	6020	0	0.2	0.00	0.54	2.78	0.02	21.68	1866	0.91
13q12dup_CRYL1	renal_failure	373542	10	7920	0	0.2	0.00	0.57	2.55	0.02	20.69	1946	0.92
13q12dup_CRYL1	thyroid	354321	9	27141	1	0.7	1.41	0.58	1.70	0.18	7.73	1948	0.93
13q12dup_CRYL1	renal	374173	10	7289	0	0.2	0.00	0.59	2.42	0.02	18.80	1991	0.93
13q12dup_CRYL1	stroke_derived2017	372644	10	8818	0	0.2	0.00	0.63	2.20	0.02	17.56	2093	0.94
13q12dup_CRYL1	hepatic	372709	10	8753	0	0.2	0.00	0.67	2.00	0.02	15.55	2194	0.95
13q12dup_CRYL1	osteoarthritis	312756	9	68706	1	2.0	0.56	0.67	0.70	0.07	3.12	2221	0.95
13q12dup_CRYL1	ovarian_cysts	374074	10	7388	0	0.2	0.00	0.71	1.82	0.01	15.01	2308	0.96
13q12dup_CRYL1	obesity	371625	10	9837	0	0.3	0.00	0.73	1.70	0.01	13.32	2390	0.96
13q12dup_CRYL1	gastrointestinal_ulcer	371110	10	10352	0	0.3	0.00	0.75	1.65	0.01	12.97	2424	0.97
13q12dup_CRYL1	connective_tissue_disorder	372110	10	9352	0	0.3	0.00	0.75	1.64	0.01	12.98	2435	0.97
13q12dup_CRYL1	MI_derived2017	367602	10	13860	0	0.4	0.00	0.77	1.58	0.01	14.02	2501	0.97
13q12dup_CRYL1	biliary	360688	10	20774	0	0.6	0.00	0.80	0.71	0.01	5.57	2589	0.97
13q12dup_CRYL1	hernia	333573	9	47889	1	1.3	0.80	0.81	1.24	0.13	5.57	2617	0.97
13q12dup_CRYL1	varicose_veins	369610	10	11852	0	0.3	0.00	0.84	1.35	0.01	10.58	2715	0.97
13q12dup_CRYL1	ear	367243	10	14219	0	0.4	0.00	0.87	1.28	0.01	9.96	2788	0.98
13q12dup_CRYL1	venous_thromboembolic_disease	367490	10	13972	0	0.4	0.00	0.87	1.27	0.01	9.91	2801	0.98
13q12dup_CRYL1	anaemia	362322	10	19140	0	0.5	0.00	0.89	0.82	0.01	6.40	2832	0.98
13q12dup_CRYL1	allergy	322801	9	58661	1	1.6	0.65	0.90	0.90	0.10	3.92	2884	0.98
13q12dup_CRYL1	ureter_bladder	332934	9	48528	1	1.3	0.79	0.91	1.11	0.12	4.87	2897	0.98
13q12dup_CRYL1	cataract	359612	10	21850	0	0.6	0.00	0.91	0.85	0.01	7.08	2901	0.98
13q12dup_CRYL1	irritable_bowel_syndrome	368459	10	13003	0	0.4	0.00	0.92	1.17	0.01	9.25	2914	0.98
13q12dup_CRYL1	COPD	367234	10	14228	0	0.4	0.00	0.92	1.15	0.01	9.32	2940	0.99
13q12dup_CRYL1	uterine_problem	339173	9	42289	1	1.1	0.90	0.93	0.92	0.10	4.37	2945	0.98
13q12dup_CRYL1	neuropathies	365559	10	15903	0	0.4	0.00	0.96	1.07	0.01	8.33	3050	0.99
13q12dup_CRYL1	diabetes_other	360728	10	20734	0	0.6	0.00	0.98	0.97	0.01	7.86	3092	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
15q11.2_del	prostate hyperplasia	368528	1523	14494	47	59.9	0.79	0.014	0.70	0.51	0.93	151	0.30
15q11.2_del	osteoarthritis	314063	1316	68959	254	289.0	0.90	0.058	0.88	0.76	1.00	358	0.50
15q11.2_del	diverticular_disease_intestine	355797	1478	27225	92	113.1	0.82	0.059	0.82	0.66	1.01	361	0.51
15q11.2_del	osteoporosis	371396	1537	11626	33	48.1	0.69	0.060	0.73	0.50	1.01	363	0.52
15q11.2_del	ischaemic heart disease not MI	366644	1515	16378	55	67.7	0.82	0.075	0.79	0.59	1.02	425	0.55
15q11.2_del	inflammatory bowel disease	364370	1508	18652	62	77.2	0.81	0.092	0.81	0.62	1.03	496	0.58
15q11.2_del	varicose veins	371133	1533	11889	37	49.1	0.76	0.11	0.77	0.55	1.05	539	0.62
15q11.2_del	irritable bowel syndrome	369977	1528	13045	42	53.9	0.78	0.14	0.80	0.58	1.07	677	0.67
15q11.2_del	migraine	369299	1525	13723	45	56.7	0.80	0.17	0.82	0.60	1.08	739	0.70
15q11.2_del	MI_derived2017	369112	1520	13910	50	57.3	0.88	0.18	0.83	0.61	1.09	771	0.72
15q11.2_del	diabetes other	362188	1470	20834	100	84.6	1.17	0.22	1.14	0.92	1.39	912	0.77
15q11.2_del	paralytic syndromes	381057	1565	1965	5	8.1	0.62	0.28	0.65	0.25	1.36	1085	0.80
15q11.2_del	ocular	361263	1471	21759	99	88.6	1.11	0.29	1.12	0.91	1.36	1134	0.81
15q11.2_del	uterine problem	340559	1395	42463	175	173.9	1.01	0.30	1.09	0.92	1.29	1171	0.81
15q11.2_del	anaemia	363795	1483	19227	87	78.4	1.10	0.31	1.12	0.90	1.38	1199	0.81
15q11.2_del	ureter bladder	334307	1382	48715	188	201.4	0.94	0.34	0.93	0.79	1.08	1271	0.83
15q11.2_del	renal failure	375063	1531	7959	39	32.5	1.20	0.34	1.17	0.84	1.59	1297	0.83
15q11.2_del	COPD	368741	1517	14281	53	58.8	0.91	0.36	0.88	0.66	1.15	1349	0.85
15q11.2_del	hepatic	374238	1539	8784	31	36.1	0.86	0.37	0.86	0.59	1.20	1386	0.85
15q11.2_del	allergy	324109	1317	58913	253	239.4	1.05	0.39	1.06	0.93	1.21	1440	0.85
15q11.2_del	asthma	333485	1357	49537	213	201.6	1.05	0.44	1.06	0.91	1.22	1567	0.87
15q11.2_del	glaucoma	376463	1539	6559	31	26.8	1.15	0.44	1.15	0.79	1.61	1594	0.87
15q11.2_del	heart valve	374616	1540	8406	30	34.6	0.87	0.45	0.87	0.60	1.23	1615	0.87
15q11.2_del	venous thromboembolic_disease	368997	1517	14025	53	57.7	0.92	0.46	0.90	0.68	1.18	1628	0.88
15q11.2_del	arrhythmia	358770	1476	24252	94	99.8	0.95	0.47	0.93	0.74	1.14	1658	0.88
15q11.2_del	died to 2016	372002	1519	11020	51	45.0	1.13	0.47	1.11	0.83	1.45	1668	0.88
15q11.2_del	low WBC count	380043	1556	2979	14	12.2	1.15	0.48	1.21	0.69	1.95	1714	0.88
15q11.2_del	diabetes insulin dependent	380415	1557	2607	13	10.7	1.22	0.51	1.21	0.67	1.98	1781	0.89
15q11.2_del	high cholesterol	316170	1298	66852	272	274.5	0.99	0.52	0.96	0.83	1.09	1804	0.90
15q11.2_del	connective tissue disorder	373629	1529	9393	41	38.4	1.07	0.55	1.10	0.79	1.48	1890	0.92
15q11.2_del	endocrine other	380651	1559	2371	11	9.7	1.13	0.56	1.20	0.63	2.04	1912	0.91
15q11.2_del	coagulation defects	380469	1558	2553	12	10.5	1.15	0.58	1.18	0.64	1.96	1950	0.93
15q11.2_del	stroke derived2017	374170	1536	8852	34	36.3	0.94	0.63	0.92	0.64	1.27	2098	0.94
15q11.2_del	obesity	373147	1532	9875	38	40.5	0.94	0.63	0.93	0.66	1.26	2106	0.94
15q11.2_del	hypertension	261880	1059	121142	511	489.9	1.03	0.65	1.03	0.92	1.15	2137	0.95
15q11.2_del	heart other	371148	1517	11874	53	48.5	1.09	0.65	1.07	0.80	1.39	2150	0.94
15q11.2_del	sciatica	339738	1397	43284	173	178.0	0.97	0.67	0.97	0.82	1.13	2208	0.95
15q11.2_del	cerebrovascular_disease_not_stroke	378676	1554	4346	16	17.8	0.90	0.70	0.91	0.54	1.43	2282	0.96
15q11.2_del	thyroid	355772	1460	27250	110	111.8	0.98	0.70	1.04	0.85	1.26	2301	0.96
15q11.2_del	digestive	300847	1239	82175	331	338.4	0.98	0.71	0.98	0.86	1.10	2309	0.96
15q11.2_del	aneurism	381047	1561	1975	9	8.1	1.11	0.71	1.13	0.55	2.02	2313	0.96
15q11.2_del	renal	375705	1542	7317	28	30.0	0.93	0.74	0.94	0.63	1.34	2416	0.97
15q11.2_del	nasal	379816	1556	3206	14	13.1	1.07	0.75	1.09	0.62	1.76	2441	0.97
15q11.2_del	ovarian cysts	375604	1540	7418	30	30.4	0.99	0.76	1.06	0.72	1.49	2468	0.97
15q11.2_del	hernia	334935	1371	48087	199	196.8	1.01	0.82	0.98	0.84	1.14	2652	0.97
15q11.2_del	biliary	362163	1485	20859	85	85.5	0.99	0.83	1.02	0.82	1.27	2673	0.97
15q11.2_del	cataract	361080	1478	21942	92	89.8	1.02	0.85	1.02	0.82	1.26	2723	0.97
15q11.2_del	respiratory	362187	1485	20835	85	85.4	1.00	0.85	0.98	0.78	1.21	2726	0.97
15q11.2_del	atherosclerotic_vascular_disease	377250	1547	5772	23	23.7	0.97	0.87	0.97	0.62	1.42	2793	0.98
15q11.2_del	ear	368743	1510	14279	60	58.5	1.03	0.87	1.02	0.78	1.31	2795	0.98
15q11.2_del	any_Ca	333126	1370	49896	200	205.2	0.98	0.88	0.99	0.85	1.15	2806	0.98
15q11.2_del	heartfailure	377925	1548	5097	22	20.9	1.05	0.91	1.02	0.65	1.52	2898	0.98
15q11.2_del	gout	375902	1540	7120	30	29.2	1.03	0.92	0.98	0.67	1.38	2915	0.98
15q11.2_del	psoriasis	376977	1545	6045	25	24.8	1.01	0.92	1.02	0.67	1.48	2923	0.98
15q11.2_del	congenital	377975	1550	5047	20	20.7	0.97	0.93	0.98	0.61	1.48	2959	0.99
15q11.2_del	gastrointestinal ulcer	372627	1527	10395	43	42.6	1.01	0.93	1.01	0.74	1.35	2962	0.99
15q11.2_del	gastric reflux	348586	1429	34436	141	141.2	1.00	0.99	1.00	0.84	1.18	3099	1.00
15q11.2_del	neuropathies	367054	1505	15968	65	65.5	0.99	0.99	1.00	0.77	1.27	3101	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
15q11.2_dup	neuropathies	367359	1810	16010	107	78.9	1.34	0.0036	1.36	1.11	1.64	70	0.16
15q11.2_dup	gout	376231	1869	7138	48	35.5	1.35	0.032	1.40	1.03	1.84	253	0.39
15q11.2_dup	atherosclerotic_vascular_disease	377579	1876	5790	41	28.8	1.42	0.035	1.42	1.03	1.91	264	0.41
15q11.2_dup	high_cholesterol	316423	1551	66946	366	328.1	1.09	0.038	1.14	1.01	1.28	279	0.43
15q11.2_dup	ischaemic_heart_disease_not_MI	366947	1818	16422	99	81.4	1.21	0.044	1.24	1.01	1.52	306	0.45
15q11.2_dup	ureter_bladder	334571	1646	48798	271	240.1	1.11	0.053	1.14	1.00	1.29	344	0.48
15q11.2_dup	ovarian_cysts	375954	1890	7415	27	37.3	0.73	0.070	0.72	0.48	1.03	401	0.54
15q11.2_dup	diverticular_disease_intestine	356120	1801	27249	116	137.8	0.85	0.086	0.85	0.70	1.02	473	0.57
15q11.2_dup	allergy	324440	1648	58929	269	299.3	0.91	0.12	0.90	0.79	1.03	588	0.63
15q11.2_dup	diabetes_insulin_dependent	380756	1898	2613	19	13.0	1.46	0.12	1.46	0.90	2.21	604	0.63
15q11.2_dup	prostate_hyperplasia	368840	1835	14529	82	72.3	1.13	0.13	1.20	0.95	1.51	636	0.64
15q11.2_dup	hypertension	262111	1290	121258	627	596.8	1.03	0.24	1.06	0.96	1.17	960	0.78
15q11.2_dup	hepatic	374564	1865	8805	52	43.8	1.18	0.26	1.18	0.88	1.53	1028	0.79
15q11.2_dup	gastric_reflux	348888	1731	34481	186	171.1	1.08	0.27	1.09	0.93	1.27	1048	0.80
15q11.2_dup	respiratory	362503	1801	20866	116	103.7	1.11	0.27	1.11	0.92	1.34	1054	0.80
15q11.2_dup	cataract	361423	1821	21946	96	110.6	0.87	0.28	0.89	0.72	1.09	1091	0.80
15q11.2_dup	obesity	373474	1859	9895	58	49.3	1.17	0.29	1.16	0.88	1.49	1124	0.81
15q11.2_dup	heartfailure	378264	1887	5105	30	25.5	1.18	0.34	1.20	0.82	1.69	1291	0.83
15q11.2_dup	COPD	369060	1836	14309	81	71.2	1.13	0.37	1.11	0.88	1.38	1383	0.85
15q11.2_dup	died_to_2016	372338	1855	11031	62	55.0	1.12	0.38	1.12	0.86	1.43	1397	0.85
15q11.2_dup	stroke_derived2017	374501	1867	8868	50	44.2	1.13	0.39	1.14	0.85	1.49	1427	0.85
15q11.2_dup	ear	369072	1839	14297	78	71.2	1.09	0.40	1.10	0.87	1.37	1484	0.85
15q11.2_dup	diabetes_other	362521	1803	20848	114	103.7	1.09	0.42	1.08	0.89	1.31	1514	0.87
15q11.2_dup	osteoporosis	371723	1864	11646	53	58.4	0.91	0.48	0.91	0.68	1.18	1698	0.88
15q11.2_dup	any_Ca	333420	1664	49949	253	249.3	1.01	0.57	1.04	0.91	1.19	1936	0.92
15q11.2_dup	migraine	369618	1844	13751	73	68.6	1.06	0.61	1.06	0.83	1.34	2032	0.94
15q11.2_dup	anaemia	364126	1814	19243	103	95.9	1.07	0.61	1.05	0.86	1.28	2041	0.94
15q11.2_dup	venous_thromboembolic_disease	369323	1843	14046	74	70.1	1.05	0.61	1.06	0.83	1.33	2049	0.94
15q11.2_dup	uterine_problem	340871	1707	42498	210	212.8	0.99	0.65	0.97	0.83	1.12	2143	0.95
15q11.2_dup	paralytic_syndromes	381398	1906	1971	11	9.8	1.12	0.67	1.14	0.60	1.93	2219	0.95
15q11.2_dup	low_WBC_count	380388	1901	2981	16	14.9	1.07	0.68	1.11	0.65	1.74	2240	0.95
15q11.2_dup	renal_failure	375412	1880	7957	37	39.8	0.93	0.68	0.93	0.66	1.27	2241	0.95
15q11.2_dup	aneurism	381395	1909	1974	8	9.9	0.81	0.69	0.87	0.41	1.61	2270	0.95
15q11.2_dup	varicose_veins	371461	1861	11908	56	59.7	0.94	0.69	0.95	0.72	1.22	2274	0.95
15q11.2_dup	digestive	301105	1497	82264	420	409.0	1.02	0.70	1.02	0.91	1.14	2289	0.95
15q11.2_dup	heart_valve	374953	1877	8416	40	42.1	0.95	0.72	0.94	0.68	1.28	2350	0.96
15q11.2_dup	biliary	362494	1816	20875	101	104.6	0.97	0.74	0.97	0.79	1.18	2403	0.96
15q11.2_dup	nasal	380163	1903	3206	14	16.0	0.87	0.75	0.92	0.52	1.49	2438	0.97
15q11.2_dup	sciatica	340035	1694	43334	223	215.9	1.03	0.76	1.02	0.89	1.17	2464	0.97
15q11.2_dup	arrhythmia	359095	1801	24274	116	121.7	0.96	0.76	0.97	0.80	1.17	2481	0.97
15q11.2_dup	inflammatory_bowel_disease	364681	1819	18688	98	93.2	1.05	0.77	1.03	0.83	1.26	2511	0.97
15q11.2_dup	connective_tissue_disorder	373972	1872	9397	45	47.0	0.96	0.78	0.96	0.70	1.27	2519	0.97
15q11.2_dup	hernia	335245	1681	48124	236	241.3	0.98	0.79	0.98	0.85	1.12	2547	0.97
15q11.2_dup	renal	376042	1879	7327	38	36.6	1.04	0.80	1.04	0.74	1.42	2581	0.97
15q11.2_dup	NI_derived2017	369438	1846	13931	71	69.6	1.02	0.82	1.03	0.80	1.30	2629	0.97
15q11.2_dup	congenital	378318	1893	5051	24	25.3	0.95	0.84	0.96	0.63	1.40	2705	0.97
15q11.2_dup	coagulation_defects	380815	1904	2554	13	12.8	1.02	0.87	1.05	0.58	1.71	2792	0.98
15q11.2_dup	endocrine_other	380997	1905	2372	12	11.9	1.01	0.87	1.05	0.57	1.75	2802	0.98
15q11.2_dup	osteoarthritis	314323	1576	69046	341	346.2	0.99	0.90	0.99	0.88	1.12	2871	0.98
15q11.2_dup	thyroid	356091	1779	27278	138	136.3	1.01	0.90	1.01	0.84	1.20	2875	0.98
15q11.2_dup	irritable_bowel_syndrome	370301	1852	13068	65	65.4	0.99	0.93	0.99	0.76	1.26	2947	0.98
15q11.2_dup	heart_other	371489	1858	11880	59	59.4	0.99	0.94	1.01	0.77	1.30	2977	0.99
15q11.2_dup	glaucoma	376810	1886	6559	31	32.8	0.94	0.94	0.99	0.68	1.38	2998	0.99
15q11.2_dup	asthma	333795	1667	49574	250	247.6	1.01	0.96	1.00	0.87	1.14	3041	0.99
15q11.2_dup	cerebrovascular_disease_not_stroke	379018	1896	4351	21	21.8	0.97	0.97	1.01	0.64	1.51	3054	0.99
15q11.2_dup	psoriasis	377319	1887	6050	30	30.3	0.99	0.98	1.00	0.68	1.40	3087	1.00
15q11.2_dup	ocular	361602	1810	21767	107	109.0	0.98	0.98	1.00	0.82	1.21	3091	1.00
15q11.2_dup	gastrointestinal_ulcer	372965	1865	10404	52	52.0	1.00	0.99	1.00	0.75	1.30	3115	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
15q11q13del_BP3-BP4	gastric_reflux	347165	8	34301	6	0.8	4.77	0.00018	9.11	3.11	25.43	23	0.025
15q11q13del_BP3-BP4	ovarian_cysts	374076	12	7390	2	0.2	7.38	0.0085	11.86	2.12	49.73	106	0.25
15q11q13del_BP3-BP4	diabetes_other	360729	11	20737	3	0.6	3.94	0.0092	6.94	1.74	21.45	116	0.25
15q11q13del_BP3-BP4	paralytic_syndromes	379505	13	1961	1	0.1	13.90	0.012	22.33	2.41	94.04	138	0.28
15q11q13del_BP3-BP4	coagulation_defects	378924	13	2542	1	0.1	10.72	0.018	17.53	1.91	71.93	172	0.32
15q11q13del_BP3-BP4	high_cholesterol	314881	9	66585	5	1.9	2.05	0.020	4.03	1.27	11.67	180	0.35
15q11q13del_BP3-BP4	varicose_veins	369612	12	11854	2	0.4	4.60	0.025	6.93	1.35	23.40	214	0.36
15q11q13del_BP3-BP4	irritable_bowel_syndrome	368461	12	13005	2	0.4	4.19	0.035	6.03	1.16	20.68	267	0.42
15q11q13del_BP3-BP4	venous_thromboembolic_disease	367492	12	13974	2	0.5	3.90	0.036	5.94	1.16	20.00	270	0.41
15q11q13del_BP3-BP4	atherosclerotic_vascular_disease	375716	13	5750	1	0.2	4.74	0.044	9.92	1.08	41.27	309	0.45
15q11q13del_BP3-BP4	ureter_bladder	332935	10	48531	4	1.5	2.25	0.050	3.40	1.00	9.82	330	0.47
15q11q13del_BP3-BP4	congenital	376438	13	5028	1	0.2	5.42	0.062	7.98	0.87	32.71	370	0.53
15q11q13del_BP3-BP4	stroke_derived2017	372647	13	8819	1	0.3	3.09	0.089	6.36	0.69	26.38	484	0.58
15q11q13del_BP3-BP4	asthma	332138	10	49328	4	1.5	2.21	0.092	2.81	0.83	8.07	497	0.58
15q11q13del_BP3-BP4	arrhythmia	357306	12	24160	2	0.8	2.26	0.099	3.81	0.74	13.00	524	0.59
15q11q13del_BP3-BP4	gastrointestinal_ulcer	371113	13	10353	1	0.4	2.63	0.13	4.98	0.54	20.77	637	0.64
15q11q13del_BP3-BP4	prostate_hyperplasia	367018	13	14448	1	0.5	1.89	0.14	5.10	0.50	27.90	675	0.67
15q11q13del_BP3-BP4	hernia	333575	11	47891	3	1.6	1.71	0.17	2.54	0.64	7.79	741	0.70
15q11q13del_BP3-BP4	obesity	371628	13	9838	1	0.3	2.77	0.17	4.17	0.45	17.34	743	0.71
15q11q13del_BP3-BP4	COPD	367237	13	14229	1	0.5	1.91	0.17	4.10	0.44	17.46	760	0.72
15q11q13del_BP3-BP4	ischaemic_heart_disease_not_MI	365142	13	16324	1	0.6	1.67	0.20	3.69	0.40	15.45	851	0.74
15q11q13del_BP3-BP4	aneurism	379500	14	1966	0	0.1	0.00	0.24	9.89	0.08	76.78	968	0.78
15q11q13del_BP3-BP4	migraine	367787	13	13679	1	0.5	1.99	0.26	3.11	0.34	13.01	1022	0.79
15q11q13del_BP3-BP4	cataract	359615	13	21851	1	0.8	1.25	0.30	2.84	0.30	12.45	1150	0.81
15q11q13del_BP3-BP4	neuropathies	365562	13	15904	1	0.6	1.71	0.31	2.71	0.29	11.15	1194	0.81
15q11q13del_BP3-BP4	hypertension	260833	12	120633	2	5.5	0.45	0.32	0.52	0.10	1.78	1232	0.82
15q11q13del_BP3-BP4	digestive	299618	10	81848	4	2.7	1.33	0.33	1.78	0.52	5.14	1249	0.82
15q11q13del_BP3-BP4	endocrine_other	379106	14	2360	0	0.1	0.00	0.33	5.92	0.05	44.57	1267	0.82
15q11q13del_BP3-BP4	diabetes_insulin_dependent	378872	14	2594	0	0.1	0.00	0.37	5.06	0.04	38.52	1367	0.85
15q11q13del_BP3-BP4	low_WBC_count	378501	14	2965	0	0.1	0.00	0.39	4.62	0.04	34.65	1447	0.85
15q11q13del_BP3-BP4	inflammatory_bowel_disease	362875	13	18591	1	0.7	1.47	0.39	2.27	0.25	9.34	1457	0.85
15q11q13del_BP3-BP4	nasal	378274	14	3192	0	0.1	0.00	0.40	4.41	0.03	33.23	1479	0.85
15q11q13del_BP3-BP4	respiratory	360715	13	20751	1	0.7	1.31	0.41	2.21	0.24	9.13	1485	0.86
15q11q13del_BP3-BP4	biliary	360691	13	20775	1	0.7	1.31	0.42	2.18	0.24	9.13	1507	0.87
15q11q13del_BP3-BP4	cerebrovascular_disease_not_stroke	377136	14	4330	0	0.2	0.00	0.42	4.15	0.03	31.64	1517	0.87
15q11q13del_BP3-BP4	ocular	359805	13	21661	1	0.8	1.26	0.42	2.15	0.23	8.89	1525	0.87
15q11q13del_BP3-BP4	heartfailure	376391	14	5075	0	0.2	0.00	0.44	3.89	0.03	29.85	1570	0.87
15q11q13del_BP3-BP4	allergy	322803	11	58663	3	2.0	1.39	0.44	1.64	0.42	4.98	1575	0.87
15q11q13del_BP3-BP4	diverticular_disease_intestine	354332	13	27134	1	1.0	1.00	0.50	1.90	0.21	8.00	1747	0.89
15q11q13del_BP3-BP4	glaucoma	374938	14	6528	0	0.2	0.00	0.58	2.49	0.02	19.18	1957	0.93
15q11q13del_BP3-BP4	gout	374376	14	7090	0	0.3	0.00	0.62	2.24	0.02	17.91	2067	0.94
15q11q13del_BP3-BP4	renal_failure	373546	14	7920	0	0.3	0.00	0.62	2.21	0.02	16.69	2077	0.94
15q11q13del_BP3-BP4	thyroid	354325	13	27141	1	1.0	1.00	0.63	1.57	0.17	6.88	2092	0.94
15q11q13del_BP3-BP4	psoriasis	375446	14	6020	0	0.2	0.00	0.63	2.16	0.02	16.23	2105	0.94
15q11q13del_BP3-BP4	renal	374177	14	7289	0	0.3	0.00	0.69	1.88	0.01	14.10	2269	0.95
15q11q13del_BP3-BP4	heart_valve	373090	14	8376	0	0.3	0.00	0.70	1.84	0.01	13.90	2291	0.95
15q11q13del_BP3-BP4	connective_tissue_disorder	372114	14	9352	0	0.4	0.00	0.76	1.61	0.01	12.28	2462	0.97
15q11q13del_BP3-BP4	hepatic	372713	14	8753	0	0.3	0.00	0.78	1.54	0.01	11.62	2524	0.97
15q11q13del_BP3-BP4	anaemia	362326	14	19140	0	0.7	0.00	0.78	0.69	0.01	5.19	2534	0.97
15q11q13del_BP3-BP4	MI_derived2017	367606	14	13860	0	0.5	0.00	0.83	1.39	0.01	10.76	2671	0.97
15q11q13del_BP3-BP4	osteoarthritis	312759	12	68707	2	2.6	0.79	0.83	1.16	0.22	4.01	2681	0.97
15q11q13del_BP3-BP4	heart_other	369645	14	11821	0	0.4	0.00	0.84	1.36	0.01	10.34	2704	0.97
15q11q13del_BP3-BP4	osteoporosis	369873	14	11593	0	0.4	0.00	0.85	1.34	0.01	10.88	2730	0.97
15q11q13del_BP3-BP4	died_to_2016	370497	14	10969	0	0.4	0.00	0.87	1.29	0.01	9.76	2778	0.98
15q11q13del_BP3-BP4	uterine_problem	339177	13	42289	1	1.6	0.64	0.88	0.87	0.09	4.16	2815	0.98
15q11q13del_BP3-BP4	any_Ca	331769	13	49697	1	1.9	0.55	0.89	0.89	0.10	3.73	2853	0.98
15q11q13del_BP3-BP4	sciatica	338354	13	43112	1	1.7	0.63	0.95	0.95	0.10	3.88	3016	0.99
15q11q13del_BP3-BP4	ear	367247	14	14219	0	0.5	0.00	1.00	1.00	0.01	7.50	3130	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
15q11q13dup_BP3_BP4	hernia	333603	39	47899	11	5.6	1.75	0.045	2.08	1.02	3.94	312	0.46
15q11q13dup_BP3_BP4	gastric_reflux	347199	42	34303	8	4.1	1.78	0.068	2.12	0.94	4.23	392	0.54
15q11q13dup_BP3_BP4	stroke_derived2017	372681	47	8821	3	1.1	2.60	0.073	3.23	0.88	8.48	417	0.55
15q11q13dup_BP3_BP4	any_Ca	331804	48	49698	2	7.2	0.31	0.081	0.37	0.08	1.11	457	0.56
15q11q13dup_BP3_BP4	ocular	359837	45	21665	5	2.7	1.76	0.12	2.19	0.80	4.93	592	0.63
15q11q13dup_BP3_BP4	inflammatory_bowel_disease	362912	50	18590	0	2.6	0.00	0.12	0.20	0.00	1.38	616	0.64
15q11q13dup_BP3_BP4	varicose_veins	369647	47	11855	3	1.5	1.93	0.15	2.48	0.68	6.42	688	0.68
15q11q13dup_BP3_BP4	heart_other	369678	47	11824	3	1.5	1.94	0.16	2.41	0.66	6.32	728	0.70
15q11q13dup_BP3_BP4	migraine	367821	47	13681	3	1.7	1.67	0.18	2.34	0.64	6.11	761	0.72
15q11q13dup_BP3_BP4	osteoarthritis	312786	39	68716	11	8.6	1.22	0.22	1.56	0.75	3.01	908	0.77
15q11q13dup_BP3_BP4	heart_valve	373124	48	8378	2	1.1	1.82	0.23	2.44	0.50	7.23	927	0.77
15q11q13dup_BP3_BP4	sciatica	338383	42	43119	8	5.4	1.42	0.23	1.61	0.71	3.20	941	0.78
15q11q13dup_BP3_BP4	MI_derived2017	367639	47	13863	3	1.8	1.65	0.26	2.01	0.54	5.37	1036	0.79
15q11q13dup_BP3_BP4	died_to_2016	370533	50	10969	0	1.5	0.00	0.30	0.31	0.00	2.14	1175	0.81
15q11q13dup_BP3_BP4	hypertension	260853	32	120649	18	14.8	1.14	0.34	1.35	0.73	2.43	1279	0.82
15q11q13dup_BP3_BP4	respiratory	360748	46	20754	4	2.6	1.47	0.34	1.65	0.54	3.94	1303	0.83
15q11q13dup_BP3_BP4	ischaemic_heart_disease_not_MI	365176	47	16326	3	2.1	1.40	0.35	1.77	0.48	4.68	1315	0.83
15q11q13dup_BP3_BP4	osteoporosis	369907	48	11595	2	1.5	1.32	0.36	1.95	0.39	6.11	1350	0.85
15q11q13dup_BP3_BP4	diabetes_other	360767	49	20735	1	2.8	0.37	0.38	0.52	0.06	1.94	1412	0.85
15q11q13dup_BP3_BP4	gastrointestinal_ulcer	371148	48	10354	2	1.3	1.47	0.40	1.83	0.38	5.40	1465	0.85
15q11q13dup_BP3_BP4	obesity	371665	50	9837	0	1.3	0.00	0.40	0.37	0.00	2.57	1480	0.85
15q11q13dup_BP3_BP4	digestive	299650	42	81852	8	11.5	0.75	0.42	0.74	0.33	1.49	1516	0.87
15q11q13dup_BP3_BP4	uterine_problem	339209	45	42293	5	5.6	0.90	0.48	1.44	0.49	3.64	1716	0.88
15q11q13dup_BP3_BP4	irritable_bowel_syndrome	368497	48	13005	2	1.7	1.17	0.49	1.63	0.33	4.81	1723	0.88
15q11q13dup_BP3_BP4	glaucoma	374973	49	6529	1	0.9	1.17	0.49	1.89	0.21	7.14	1726	0.89
15q11q13dup_BP3_BP4	anaemia	362361	49	19141	1	2.6	0.40	0.50	0.60	0.07	2.22	1769	0.89
15q11q13dup_BP3_BP4	biliary	360725	47	20777	3	2.7	1.10	0.52	1.47	0.40	3.83	1806	0.90
15q11q13dup_BP3_BP4	diverticular_disease_intestine	354365	46	27137	4	3.5	1.12	0.52	1.40	0.45	3.40	1814	0.90
15q11q13dup_BP3_BP4	venous_thromboembolic_disease	367528	48	13974	2	1.8	1.09	0.59	1.45	0.30	4.29	1977	0.93
15q11q13dup_BP3_BP4	renal	374213	50	7289	0	1.0	0.00	0.59	0.51	0.00	3.54	1994	0.93
15q11q13dup_BP3_BP4	ureter_bladder	332968	43	48534	7	6.3	1.10	0.61	1.23	0.52	2.54	2031	0.94
15q11q13dup_BP3_BP4	aneurism	379536	50	1966	0	0.3	0.00	0.66	2.02	0.02	14.35	2176	0.95
15q11q13dup_BP3_BP4	psoriasis	375482	50	6020	0	0.8	0.00	0.66	0.57	0.00	3.95	2180	0.95
15q11q13dup_BP3_BP4	renal_failure	373581	49	7921	1	1.0	0.96	0.67	1.46	0.16	5.48	2211	0.95
15q11q13dup_BP3_BP4	gout	374411	49	7091	1	0.9	1.08	0.70	1.41	0.16	5.36	2283	0.96
15q11q13dup_BP3_BP4	neuropathies	365597	48	15905	2	2.1	0.96	0.70	1.30	0.27	3.83	2290	0.95
15q11q13dup_BP3_BP4	endocrine_other	379142	50	2360	0	0.3	0.00	0.71	1.77	0.01	12.38	2316	0.96
15q11q13dup_BP3_BP4	paralytic_syndromes	379542	50	1960	0	0.3	0.00	0.72	1.73	0.01	12.10	2356	0.96
15q11q13dup_BP3_BP4	connective_tissue_disorder	372149	49	9353	1	1.2	0.82	0.73	1.35	0.15	5.06	2383	0.96
15q11q13dup_BP3_BP4	asthma	332171	43	49331	7	6.4	1.08	0.74	1.14	0.48	2.34	2407	0.96
15q11q13dup_BP3_BP4	atherosclerotic_vascular_disease	375753	50	5749	0	0.8	0.00	0.75	0.66	0.01	4.61	2433	0.97
15q11q13dup_BP3_BP4	prostate_hyperplasia	367054	49	14448	1	1.9	0.53	0.77	0.78	0.09	3.16	2483	0.97
15q11q13dup_BP3_BP4	thyroid	354359	47	27143	3	3.6	0.84	0.77	1.19	0.32	3.18	2485	0.97
15q11q13dup_BP3_BP4	ovarian_cysts	374114	50	7388	0	1.0	0.00	0.78	0.68	0.01	4.98	2515	0.97
15q11q13dup_BP3_BP4	ear	367282	49	14220	1	1.9	0.54	0.78	0.80	0.09	2.96	2529	0.97
15q11q13dup_BP3_BP4	coagulation_defects	378961	50	2541	0	0.3	0.00	0.80	1.45	0.01	10.12	2595	0.97
15q11q13dup_BP3_BP4	hepatic	372748	49	8754	1	1.2	0.87	0.80	1.24	0.14	4.57	2598	0.97
15q11q13dup_BP3_BP4	COPD	367273	49	14229	1	1.9	0.54	0.81	0.82	0.09	3.09	2602	0.97
15q11q13dup_BP3_BP4	arrhythmia	357341	47	24161	3	3.2	0.95	0.81	1.15	0.31	3.00	2613	0.97
15q11q13dup_BP3_BP4	congenital	376475	50	5027	0	0.7	0.00	0.81	0.73	0.01	5.04	2615	0.97
15q11q13dup_BP3_BP4	heartfailure	376427	50	5075	0	0.7	0.00	0.84	0.76	0.01	5.38	2707	0.97
15q11q13dup_BP3_BP4	low_WBC_count	378537	50	2965	0	0.4	0.00	0.84	1.35	0.01	9.41	2710	0.97
15q11q13dup_BP3_BP4	diabetes_insulin_dependent	378908	50	2594	0	0.3	0.00	0.85	1.32	0.01	9.21	2744	0.97
15q11q13dup_BP3_BP4	cataract	359650	48	21852	2	2.9	0.70	0.90	0.92	0.19	2.82	2864	0.98
15q11q13dup_BP3_BP4	nasal	378310	50	3192	0	0.4	0.00	0.91	1.19	0.01	8.27	2889	0.98
15q11q13dup_BP3_BP4	cerebrovascular_disease_not_stroke	377172	50	4330	0	0.6	0.00	0.95	0.92	0.01	6.48	3015	0.99
15q11q13dup_BP3_BP4	allergy	322835	43	58667	7	7.8	0.91	0.99	0.99	0.42	2.04	3107	1.00
15q11q13dup_BP3_BP4	high_cholesterol	314914	42	66588	8	8.9	0.92	0.99	1.00	0.44	2.07	3110	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
15q11q13dup_BP3_BP5	ischaemic_heart_disease_not_MI	365136	7	16325	2	0.3	5.19	0.013	10.30	1.80	43.33	141	0.28
15q11q13dup_BP3_BP5	congenital	376433	8	5028	1	0.1	8.43	0.035	11.81	1.26	52.52	265	0.41
15q11q13dup_BP3_BP5	renal	374171	8	7290	1	0.2	5.81	0.053	9.12	0.97	41.02	341	0.49
15q11q13dup_BP3_BP5	thyroid	354319	7	27142	2	0.5	3.12	0.079	4.66	0.81	19.89	448	0.55
15q11q13dup_BP3_BP5	died_to_2016	370491	8	10970	1	0.2	3.86	0.11	5.75	0.60	26.42	551	0.62
15q11q13dup_BP3_BP5	asthma	332134	6	49327	3	0.9	2.58	0.11	3.15	0.75	11.12	552	0.62
15q11q13dup_BP3_BP5	osteoarthritis	312753	6	68708	3	1.3	1.85	0.15	2.95	0.65	11.50	682	0.68
15q11q13dup_BP3_BP5	aneurism	379495	9	1966	0	0.0	0.00	0.19	14.74	0.11	126.55	807	0.74
15q11q13dup_BP3_BP5	neuropathies	365557	8	15904	1	0.3	2.67	0.20	3.85	0.41	17.37	824	0.74
15q11q13dup_BP3_BP5	hypertension	260826	5	120635	4	2.3	1.41	0.23	2.31	0.58	8.86	923	0.77
15q11q13dup_BP3_BP5	inflammatory_bowel_disease	362870	8	18591	1	0.4	2.28	0.24	3.31	0.35	14.81	970	0.78
15q11q13dup_BP3_BP5	diabetes_other	360726	8	20735	1	0.5	2.04	0.25	3.32	0.34	15.84	997	0.78
15q11q13dup_BP3_BP5	paralytic_syndromes	379501	9	1960	0	0.0	0.00	0.26	9.08	0.07	73.83	1020	0.79
15q11q13dup_BP3_BP5	allergy	322801	9	58660	0	1.6	0.00	0.27	0.27	0.00	2.16	1069	0.80
15q11q13dup_BP3_BP5	endocrine_other	379101	9	2360	0	0.1	0.00	0.28	8.02	0.06	64.34	1083	0.80
15q11q13dup_BP3_BP5	coagulation_defects	378920	9	2541	0	0.1	0.00	0.28	7.68	0.06	60.77	1113	0.80
15q11q13dup_BP3_BP5	nasal	378269	9	3192	0	0.1	0.00	0.30	7.15	0.06	57.15	1147	0.81
15q11q13dup_BP3_BP5	low_WBC_count	378496	9	2965	0	0.1	0.00	0.30	7.14	0.06	57.03	1149	0.81
15q11q13dup_BP3_BP5	digestive	299614	6	81847	3	1.6	1.55	0.30	2.08	0.48	7.55	1158	0.81
15q11q13dup_BP3_BP5	diabetes_insulin_dependent	378867	9	2594	0	0.1	0.00	0.32	6.42	0.05	52.48	1228	0.82
15q11q13dup_BP3_BP5	cerebrovascular_disease_not_stroke	377131	9	4330	0	0.1	0.00	0.36	5.44	0.04	46.94	1336	0.84
15q11q13dup_BP3_BP5	heartfailure	376386	9	5075	0	0.1	0.00	0.39	4.73	0.04	40.87	1443	0.85
15q11q13dup_BP3_BP5	glaucoma	374933	9	6528	0	0.2	0.00	0.43	4.00	0.03	33.64	1557	0.87
15q11q13dup_BP3_BP5	sciatica	338350	9	43111	0	1.1	0.00	0.45	0.39	0.00	3.09	1620	0.88
15q11q13dup_BP3_BP5	atherosclerotic_vascular_disease	375712	9	5749	0	0.1	0.00	0.46	3.60	0.03	29.69	1646	0.88
15q11q13dup_BP3_BP5	ureter_bladder	332934	9	48527	0	1.3	0.00	0.47	0.40	0.00	3.20	1655	0.88
15q11q13dup_BP3_BP5	gout	374371	9	7090	0	0.2	0.00	0.48	3.50	0.03	32.38	1685	0.88
15q11q13dup_BP3_BP5	prostate_hyperplasia	367014	9	14447	0	0.4	0.00	0.52	3.08	0.02	37.19	1827	0.90
15q11q13dup_BP3_BP5	psoriasis	375441	9	6020	0	0.1	0.00	0.53	2.88	0.02	22.88	1842	0.90
15q11q13dup_BP3_BP5	renal_failure	373541	9	7920	0	0.2	0.00	0.54	2.83	0.02	23.65	1854	0.91
15q11q13dup_BP3_BP5	heart_valve	373085	9	8376	0	0.2	0.00	0.56	2.65	0.02	21.69	1919	0.91
15q11q13dup_BP3_BP5	stroke_derived2017	372643	9	8818	0	0.2	0.00	0.60	2.39	0.02	19.87	2009	0.93
15q11q13dup_BP3_BP5	ovarian_cysts	374073	9	7388	0	0.2	0.00	0.63	2.24	0.02	19.98	2078	0.94
15q11q13dup_BP3_BP5	gastric_reflux	347166	9	34295	0	0.9	0.00	0.64	0.54	0.00	4.32	2126	0.95
15q11q13dup_BP3_BP5	connective_tissue_disorder	372109	9	9352	0	0.2	0.00	0.65	2.10	0.02	17.28	2151	0.94
15q11q13dup_BP3_BP5	hepatic	372708	9	8753	0	0.2	0.00	0.66	2.05	0.02	16.37	2169	0.95
15q11q13dup_BP3_BP5	heart_other	369640	9	11821	0	0.3	0.00	0.67	2.01	0.02	16.65	2196	0.95
15q11q13dup_BP3_BP5	any_Ca	331764	8	49697	1	1.2	0.85	0.67	1.51	0.15	7.22	2198	0.95
15q11q13dup_BP3_BP5	gastrointestinal_ulcer	371109	9	10352	0	0.3	0.00	0.68	1.93	0.01	15.89	2245	0.95
15q11q13dup_BP3_BP5	hernia	333572	8	47889	1	1.1	0.89	0.72	1.40	0.15	6.50	2354	0.96
15q11q13dup_BP3_BP5	MI_derived2017	367601	9	13860	0	0.3	0.00	0.73	1.73	0.01	15.30	2389	0.96
15q11q13dup_BP3_BP5	varicose_veins	369609	9	11852	0	0.3	0.00	0.74	1.67	0.01	13.38	2414	0.97
15q11q13dup_BP3_BP5	obesity	371624	9	9837	0	0.2	0.00	0.77	1.57	0.01	12.61	2492	0.97
15q11q13dup_BP3_BP5	osteoporosis	369868	9	11593	0	0.3	0.00	0.82	1.44	0.01	14.44	2646	0.97
15q11q13dup_BP3_BP5	venous_thromboembolic_disease	367489	9	13972	0	0.3	0.00	0.82	1.41	0.01	11.37	2651	0.97
15q11q13dup_BP3_BP5	irritable_bowel_syndrome	368458	9	13003	0	0.3	0.00	0.83	1.38	0.01	11.11	2676	0.97
15q11q13dup_BP3_BP5	diverticular_disease_intestine	354328	9	27133	0	0.7	0.00	0.84	0.75	0.01	6.54	2708	0.97
15q11q13dup_BP3_BP5	uterine_problem	339172	8	42289	1	1.0	1.00	0.84	1.21	0.12	6.57	2709	0.97
15q11q13dup_BP3_BP5	ear	367242	9	14219	0	0.3	0.00	0.84	1.35	0.01	10.85	2716	0.97
15q11q13dup_BP3_BP5	COPD	367233	9	14228	0	0.3	0.00	0.85	1.33	0.01	11.03	2750	0.97
15q11q13dup_BP3_BP5	migraine	367783	9	13678	0	0.3	0.00	0.87	1.29	0.01	10.50	2780	0.98
15q11q13dup_BP3_BP5	respiratory	360711	9	20750	0	0.5	0.00	0.94	0.90	0.01	7.29	2993	0.99
15q11q13dup_BP3_BP5	biliary	360687	9	20774	0	0.5	0.00	0.95	0.91	0.01	7.40	3000	0.99
15q11q13dup_BP3_BP5	cataract	359611	9	21850	0	0.5	0.00	0.95	1.10	0.01	10.14	3010	0.99
15q11q13dup_BP3_BP5	anaemia	362321	9	19140	0	0.5	0.00	0.96	0.93	0.01	7.41	3032	0.99
15q11q13dup_BP3_BP5	arrhythmia	357303	9	24158	0	0.6	0.00	0.97	0.94	0.01	7.81	3059	0.99
15q11q13dup_BP3_BP5	ocular	359801	9	21660	0	0.5	0.00	0.99	1.02	0.01	8.26	3109	1.00
15q11q13dup_BP3_BP5	high_cholesterol	314880	8	66581	1	1.7	0.64	1.00	1.00	0.10	4.94	3128	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
15q13.3_del	asthma	332151	23	49338	14	3.4	2.93	0.00018	3.90	1.97	7.43	22	0.026
15q13.3_del	diabetes_other	360746	28	20743	9	1.6	4.48	0.00038	4.87	2.15	10.15	31	0.039
15q13.3_del	COPD	367255	31	14234	6	1.2	4.35	0.0093	3.84	1.44	8.82	120	0.24
15q13.3_del	ocular	359823	31	21666	6	1.9	2.86	0.012	3.52	1.37	7.75	136	0.27
15q13.3_del	high_cholesterol	314897	25	66592	12	5.3	1.86	0.042	2.20	1.03	4.48	297	0.44
15q13.3_del	inflammatory_bowel_disease	362894	32	18595	5	1.6	2.77	0.043	2.89	1.04	6.63	301	0.45
15q13.3_del	hypertension	260840	19	120649	18	8.8	1.54	0.050	1.98	1.00	3.92	328	0.47
15q13.3_del	neuropathies	365582	33	15907	4	1.4	2.59	0.067	2.87	0.92	6.99	387	0.54
15q13.3_del	any_Ca	331792	36	49697	1	5.4	0.21	0.075	0.29	0.03	1.11	430	0.55
15q13.3_del	endocrine_other	379128	36	2361	1	0.2	4.37	0.088	6.22	0.70	23.42	481	0.57
15q13.3_del	obesity	371649	34	9840	3	0.9	3.14	0.11	2.83	0.76	7.52	543	0.62
15q13.3_del	irritable_bowel_syndrome	368483	34	13006	3	1.2	2.38	0.11	2.81	0.76	7.49	553	0.62
15q13.3_del	died_to_2016	370517	34	10972	3	1.0	2.82	0.13	2.63	0.71	7.02	646	0.64
15q13.3_del	diabetes_insulin_dependent	378894	36	2595	1	0.2	3.97	0.15	4.42	0.50	16.70	681	0.68
15q13.3_del	osteoarthritis	312774	27	68715	10	5.9	1.50	0.15	1.74	0.80	3.53	699	0.69
15q13.3_del	gout	374397	35	7092	2	0.7	2.91	0.18	2.82	0.56	8.91	769	0.72
15q13.3_del	osteoporosis	369894	35	11595	2	1.1	1.78	0.18	2.81	0.56	8.80	774	0.72
15q13.3_del	renal_failure	373567	35	7922	2	0.7	2.60	0.20	2.64	0.53	8.10	832	0.75
15q13.3_del	allergy	322821	29	58668	8	5.3	1.41	0.20	1.69	0.74	3.49	841	0.74
15q13.3_del	prostate_hyperplasia	367039	34	14450	3	1.3	2.14	0.20	2.32	0.59	6.86	849	0.74
15q13.3_del	cataract	359636	34	21853	3	2.1	1.42	0.33	1.83	0.49	5.01	1255	0.82
15q13.3_del	migraine	367809	35	13680	2	1.3	1.51	0.34	2.01	0.41	6.12	1276	0.83
15q13.3_del	biliary	360712	34	20777	3	2.0	1.49	0.36	1.74	0.47	4.63	1342	0.84
15q13.3_del	digestive	299634	26	81855	11	7.1	1.39	0.36	1.40	0.66	2.76	1346	0.85
15q13.3_del	diverticular_disease_intestine	354355	36	27134	1	2.8	0.38	0.39	0.52	0.06	1.97	1426	0.85
15q13.3_del	gastrointestinal_ulcer	371135	35	10354	2	1.0	1.99	0.39	1.86	0.37	5.73	1455	0.85
15q13.3_del	anaemia	362346	34	19143	3	1.8	1.62	0.40	1.66	0.45	4.39	1475	0.85
15q13.3_del	psoriasis	375468	36	6021	1	0.6	1.71	0.40	2.20	0.25	8.25	1477	0.85
15q13.3_del	varicose_veins	369637	37	11850	0	1.2	0.00	0.44	0.39	0.00	2.78	1591	0.87
15q13.3_del	atherosclerotic_vascular_disease	375739	36	5750	1	0.6	1.79	0.46	2.00	0.22	7.65	1630	0.88
15q13.3_del	venous_thromboembolic_disease	367515	35	13974	2	1.3							

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
15q13.3_dup	renal	374375	212	7301	12	4.1	2.80	0.0017	2.89	1.55	4.91	49	0.11
15q13.3_dup	died_to_2016	370690	207	10986	17	6.1	2.64	0.0017	2.42	1.43	3.85	50	0.11
15q13.3_dup	stroke_derived2017	372846	212	8830	12	5.0	2.32	0.015	2.23	1.19	3.81	156	0.30
15q13.3_dup	congenital	376642	217	5034	7	2.9	2.37	0.035	2.44	1.07	4.70	269	0.41
15q13.3_dup	prostate_hyperplasia	367225	220	14451	4	8.7	0.47	0.040	0.42	0.14	0.97	294	0.43
15q13.3_dup	asthma	332332	204	49344	20	30.3	0.69	0.047	0.65	0.40	0.99	318	0.46
15q13.3_dup	migraine	367996	222	13680	2	8.3	0.25	0.048	0.35	0.07	0.99	321	0.47
15q13.3_dup	obesity	371828	213	9848	11	5.6	1.90	0.081	1.78	0.93	3.09	453	0.56
15q13.3_dup	renal_failure	373747	215	7929	9	4.6	1.94	0.085	1.88	0.91	3.43	471	0.57
15q13.3_dup	COPD	367433	209	14243	15	8.1	1.80	0.10	1.61	0.91	2.65	530	0.60
15q13.3_dup	ischaemic_heart_disease_not_MI	365338	209	16338	15	9.3	1.56	0.12	1.55	0.88	2.55	605	0.63
15q13.3_dup	endocrine_other	379313	221	2363	3	1.4	2.16	0.12	2.62	0.73	6.50	608	0.63
15q13.3_dup	anaemia	362520	208	19156	16	11.0	1.42	0.14	1.49	0.87	2.39	671	0.66
15q13.3_dup	diverticular_disease_intestine	354533	214	27143	10	16.4	0.63	0.17	0.66	0.33	1.17	740	0.70
15q13.3_dup	nasal	378484	224	3192	0	1.9	0.00	0.22	0.26	0.00	1.75	894	0.76
15q13.3_dup	biliary	360887	209	20789	15	12.0	1.23	0.23	1.40	0.80	2.28	922	0.77
15q13.3_dup	ocular	360008	216	21668	8	13.0	0.63	0.23	0.67	0.31	1.26	932	0.77
15q13.3_dup	arrhythmia	357499	205	24177	19	13.9	1.34	0.23	1.35	0.82	2.10	934	0.78
15q13.3_dup	osteoarthritis	312939	192	68737	32	42.2	0.79	0.26	0.81	0.54	1.17	1040	0.80
15q13.3_dup	high_cholesterol	315061	189	66615	35	40.0	0.90	0.27	0.81	0.55	1.17	1068	0.80
15q13.3_dup	allergy	322989	197	58687	27	35.8	0.78	0.28	0.81	0.53	1.18	1079	0.80
15q13.3_dup	hepatic	372915	216	8761	8	5.1	1.56	0.29	1.48	0.69	2.76	1132	0.81
15q13.3_dup	hypertension	260965	144	120711	80	66.6	1.13	0.30	1.16	0.87	1.55	1152	0.81
15q13.3_dup	coagulation_defects	379135	224	2541	0	1.5	0.00	0.31	0.31	0.00	2.11	1185	0.81
15q13.3_dup	uterine_problem	339374	210	42302	14	26.2	0.56	0.33	0.76	0.42	1.30	1258	0.82
15q13.3_dup	hernia	333763	199	47913	25	28.6	0.89	0.33	0.82	0.53	1.22	1259	0.82
15q13.3_dup	connective_tissue_disorder	372317	217	9359	7	5.5	1.27	0.33	1.47	0.64	2.84	1266	0.82
15q13.3_dup	ear	367446	213	14230	11	8.2	1.32	0.34	1.36	0.71	2.35	1282	0.82
15q13.3_dup	heartfailure	376596	219	5080	5	3.0	1.68	0.34	1.56	0.58	3.34	1285	0.82
15q13.3_dup	osteoporosis	370080	221	11596	3	6.9	0.44	0.35	0.63	0.17	1.57	1324	0.84
15q13.3_dup	gastric_reflux	347365											



CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
15q13.3del(CHRNA7)	heart_valve	373084	8	8378	2	0.2	9.11	0.0046	14.45	2.69	53.98	77	0.19
15q13.3del(CHRNA7)	diabetes_insulin_dependent	378867	9	2595	1	0.1	14.71	0.021	16.19	1.70	72.94	195	0.35
15q13.3del(CHRNA7)	ischaemic_heart_disease_not_MI	365137	8	16325	2	0.4	4.67	0.027	7.22	1.31	28.17	228	0.37
15q13.3del(CHRNA7)	asthma	332134	6	49328	4	0.9	3.09	0.031	4.21	1.16	13.95	245	0.39
15q13.3del(CHRNA7)	inflammatory_bowel_disease	362870	8	18592	2	0.4	4.10	0.045	5.54	1.04	20.23	313	0.46
15q13.3del(CHRNA7)	ureter_bladder	332932	7	48530	3	1.0	2.36	0.097	3.28	0.78	11.27	517	0.59
15q13.3del(CHRNA7)	gout	374371	9	7091	1	0.2	5.38	0.099	6.18	0.64	29.22	525	0.59
15q13.3del(CHRNA7)	stroke_derived2017	372643	9	8819	1	0.2	4.33	0.11	5.82	0.61	26.91	547	0.62
15q13.3del(CHRNA7)	allergy	322799	7	58663	3	1.3	1.95	0.13	2.86	0.69	9.70	651	0.65
15q13.3del(CHRNA7)	obesity	371624	9	9838	1	0.2	3.88	0.14	4.77	0.51	21.20	673	0.66
15q13.3del(CHRNA7)	heart_other	369640	9	11822	1	0.3	3.23	0.15	4.64	0.49	21.17	691	0.68
15q13.3del(CHRNA7)	osteoarthritis	312754	7	68708	3	1.5	1.67	0.16	2.79	0.65	9.86	705	0.69
15q13.3del(CHRNA7)	ear	367242	9	14220	1	0.3	2.68	0.17	4.17	0.44	18.25	753	0.72
15q13.3del(CHRNA7)	neuropathies	365558	9	15904	1	0.4	2.40	0.19	3.86	0.41	16.96	814	0.74
15q13.3del(CHRNA7)	high_cholesterol	314879	7	66583	3	1.5	1.72	0.22	2.41	0.56	8.78	904	0.76
15q13.3del(CHRNA7)	biliary	360687	9	20775	1	0.5	1.84	0.24	3.30	0.35	14.68	973	0.78
15q13.3del(CHRNA7)	COPD	367233	9	14229	1	0.3	2.68	0.26	3.23	0.33	15.36	1024	0.79
15q13.3del(CHRNA7)	aneurism	379496	10	1966	0	0.1	0.00	0.26	8.75	0.07	72.00	1035	0.79
15q13.3del(CHRNA7)	endocrine_other	379102	10	2360	0	0.1	0.00	0.27	8.51	0.07	66.77	1044	0.80
15q13.3del(CHRNA7)	ocular	359801	9	21661	1	0.5	1.76	0.27	3.05	0.32	13.51	1059	0.80
15q13.3del(CHRNA7)	sciatica	338349	8	43113	2	1.0	1.77	0.29	2.29	0.43	8.34	1125	0.81
15q13.3del(CHRNA7)	low_WBC_count	378497	10	2965	0	0.1	0.00	0.31	6.71	0.05	52.32	1188	0.81
15q13.3del(CHRNA7)	paralytic_syndromes	379502	10	1960	0	0.1	0.00	0.32	6.52	0.05	52.45	1214	0.81
15q13.3del(CHRNA7)	coagulation_defects	378921	10	2541	0	0.1	0.00	0.32	6.37	0.05	49.77	1229	0.81
15q13.3del(CHRNA7)	nasal	378270	10	3192	0	0.1	0.00	0.33	6.17	0.05	48.35	1245	0.82
15q13.3del(CHRNA7)	arrhythmia	357303	9	24159	1	0.6	1.58	0.37	2.44	0.26	10.99	1358	0.84
15q13.3del(CHRNA7)	cerebrovascular_disease_not_stroke	377132	10	4330	0	0.1	0.00	0.38	4.89	0.04	39.47	1405	0.85
15q13.3del(CHRNA7)	any_Ca	331766	10	49696	0	1.5	0.00	0.45	0.39	0.00	3.07	1607	0.87
15q13.3del(CHRNA7)	uterine_problem	339173	9	42289	1	1.1	0.90	0.48	2.15	0.20	16.23	1693	0.88
15q13.3del(CHRNA7)	glaucoma	374934	10	6528	0	0.2	0.00	0.48	3.40	0.03	27.85	1701	0.88
15q13.3del(CHRNA7)	ovarian_cysts	374074	10	7388	0	0.2	0.00	0.49	3.34	0.02	34.92	1737	0.89
15q13.3del(CHRNA7)	hypertension	260827	6	120635	4	2.8	1.26	0.50	1.55	0.41	5.43	1760	0.89
15q13.3del(CHRNA7)	congenital	376435	10	5027	0	0.1	0.00	0.50	3.14	0.02	24.43	1764	0.89
15q13.3del(CHRNA7)	heartfailure	376387	10	5075	0	0.1	0.00	0.54	2.82	0.02	23.68	1859	0.91
15q13.3del(CHRNA7)	atherosclerotic_vascular_disease	375713	10	5749	0	0.2	0.00	0.57	2.55	0.02	20.84	1944	0.92
15q13.3del(CHRNA7)	psoriasis	375442	10	6020	0	0.2	0.00	0.59	2.45	0.02	19.20	1974	0.93
15q13.3del(CHRNA7)	gastric_reflux	347167	10	34295	0	1.0	0.00	0.59	0.49	0.00	3.87	1979	0.93
15q13.3del(CHRNA7)	osteoporosis	369869	10	11593	0	0.3	0.00	0.59	2.47	0.02	24.07	1998	0.93
15q13.3del(CHRNA7)	renal	374173	10	7289	0	0.2	0.00	0.61	2.27	0.02	17.78	2055	0.94
15q13.3del(CHRNA7)	connective_tissue_disorder	372110	10	9352	0	0.3	0.00	0.62	2.22	0.02	17.79	2074	0.94
15q13.3del(CHRNA7)	renal_failure	373542	10	7920	0	0.2	0.00	0.68	1.95	0.01	15.94	2230	0.95
15q13.3del(CHRNA7)	varicose_veins	369610	10	11852	0	0.3	0.00	0.75	1.66	0.01	13.03	2417	0.97
15q13.3del(CHRNA7)	diabetes_other	360728	10	20734	0	0.6	0.00	0.75	0.65	0.00	5.29	2436	0.97
15q13.3del(CHRNA7)	hepatic	372709	10	8753	0	0.2	0.00	0.75	1.63	0.01	12.89	2446	0.96
15q13.3del(CHRNA7)	respiratory	360712	10	20750	0	0.6	0.00	0.77	0.67	0.01	5.37	2502	0.97
15q13.3del(CHRNA7)	migraine	367784	10	13678	0	0.4	0.00	0.78	1.55	0.01	12.72	2526	0.97
15q13.3del(CHRNA7)	irritable_bowel_syndrome	368459	10	13003	0	0.4	0.00	0.78	1.53	0.01	12.27	2537	0.97
15q13.3del(CHRNA7)	gastrointestinal_ulcer	371110	10	10352	0	0.3	0.00	0.81	1.43	0.01	11.54	2625	0.97
15q13.3del(CHRNA7)	diverticular_disease_intestine	354329	10	27133	0	0.8	0.00	0.83	0.74	0.01	5.99	2674	0.97
15q13.3del(CHRNA7)	prostate_hyperplasia	367015	10	14447	0	0.4	0.00	0.86	1.32	0.01	11.36	2759	0.97
15q13.3del(CHRNA7)	venous_thromboembolic_disease	367490	10	13972	0	0.4	0.00	0.87	1.27	0.01	10.00	2800	0.98
15q13.3del(CHRNA7)	died_to_2016	370493	10	10969	0	0.3	0.00	0.92	1.17	0.01	9.32	2922	0.98
15q13.3del(CHRNA7)	anaemia	362322	10	19140	0	0.5	0.00	0.93	0.89	0.01	6.97	2960	0.99
15q13.3del(CHRNA7)	thyroid	354322	10	27140	0	0.8	0.00	0.94	0.89	0.01	7.61	2970	0.99
15q13.3del(CHRNA7)	digestive	299616	8	81846	2	2.2	0.93	0.96	1.04	0.19	3.82	3033	0.99
15q13.3del(CHRNA7)	cataract	359612	10	21850	0	0.6	0.00	0.97	1.06	0.01	9.26	3055	0.99
15q13.3del(CHRNA7)	hernia	333573	9	47889	1	1.3	0.80	0.98	1.02	0.11	4.56	3089	1.00
15q13.3del(CHRNA7)	MI_derived2017	367602	10	13860	0	0.4	0.00	0.99	0.98	0.01	8.20	3102	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
15q13.3dup(CHRNA7)	heart_valve	375839	2763	8456	80	62.2	1.28	0.020	1.32	1.05	1.64	177	0.35
15q13.3dup(CHRNA7)	allergy	325161	2369	59134	474	430.8	1.08	0.047	1.11	1.00	1.22	320	0.46
15q13.3dup(CHRNA7)	high_cholesterol	317265	2393	67030	450	505.6	0.91	0.069	0.91	0.82	1.01	397	0.55
15q13.3dup(CHRNA7)	cerebrovascular_disease_not_stroke	379925	2803	4370	40	32.2	1.24	0.12	1.30	0.93	1.75	591	0.63
15q13.3dup(CHRNA7)	biliary	363349	2671	20946	172	154.0	1.11	0.13	1.13	0.96	1.32	645	0.64
15q13.3dup(CHRNA7)	gastric_reflux	349772	2615	34523	228	258.1	0.89	0.14	0.90	0.79	1.03	657	0.65
15q13.3dup(CHRNA7)	inflammatory_bowel_disease	365551	2689	18744	154	137.9	1.11	0.15	1.13	0.96	1.32	698	0.68
15q13.3dup(CHRNA7)	gout	377143	2781	7152	62	52.7	1.17	0.16	1.21	0.93	1.55	709	0.69
15q13.3dup(CHRNA7)	thyroid	356935	2623	27360	220	201.1	1.09	0.17	1.10	0.96	1.27	748	0.71
15q13.3dup(CHRNA7)	heart_other	372375	2744	11920	99	87.8	1.12	0.18	1.15	0.94	1.40	762	0.72
15q13.3dup(CHRNA7)	diverticular_disease_intestine	356982	2663	27313	180	203.7	0.89	0.19	0.90	0.77	1.05	804	0.73
15q13.3dup(CHRNA7)	low_WBC_count	381302	2815	2993	28	22.1	1.27	0.19	1.29	0.87	1.84	813	0.74
15q13.3dup(CHRNA7)	ischaemic_heart_disease_not_MI	367841	2712	16454	131	121.3	1.08	0.22	1.12	0.93	1.33	895	0.76
15q13.3dup(CHRNA7)	hypertension	262751	1930	121544	913	892.8	1.02	0.25	1.05	0.97	1.14	1002	0.78
15q13.3dup(CHRNA7)	renal	376944	2781	7351	62	54.2	1.14	0.26	1.16	0.89	1.48	1018	0.79
15q13.3dup(CHRNA7)	MI_derived2017	370345	2753	13950	90	103.7	0.87	0.27	0.89	0.71	1.09	1053	0.80
15q13.3dup(CHRNA7)	sciatica	340850	2509	43445	334	319.8	1.04	0.29	1.07	0.95	1.19	1116	0.80
15q13.3dup(CHRNA7)	congenital	379237	2812	5058	31	37.5	0.83	0.31	0.84	0.58	1.17	1211	0.81
15q13.3dup(CHRNA7)	arrhythmia	359949	2655	24346	188	179.6	1.04	0.37	1.07	0.92	1.24	1365	0.85
15q13.3dup(CHRNA7)	digestive	301868	2260	82427	583	617.1	0.96	0.37	0.96	0.87	1.05	1377	0.85
15q13.3dup(CHRNA7)	obesity	374379	2764	9916	79	73.2	1.08	0.38	1.11	0.88	1.38	1410	0.85
15q13.3dup(CHRNA7)	diabetes_other	363398	2680	20897	163	154.1	1.05	0.39	1.07	0.91	1.26	1432	0.85
15q13.3dup(CHRNA7)	ocular	362486	2694	21809	149	162.1	0.92	0.41	0.93	0.79	1.10	1501	0.86
15q13.3dup(CHRNA7)	COPD	369972	2748	14323	95	106.4	0.90	0.46	0.93	0.75	1.13	1639	0.88
15q13.3dup(CHRNA7)	neuropathies	368268	2719	16027	124	118.3	1.05	0.50	1.06	0.88	1.27	1763	0.89
15q13.3dup(CHRNA7)	osteoarthritis	315078	2331	69217	512	512.1	1.00	0.54	1.03	0.93	1.14	1852	0.91
15q13.3dup(CHRNA7)	glaucoma	377724	2800	6571	43	48.7	0.88	0.54	0.91	0.66	1.22	1856	0.91
15q13.3dup(CHRNA7)	ovarian_cysts	376856	2792	7439	51	55.1	0.93	0.56	0.92	0.69	1.20	1898	0.92
15q13.3dup(CHRNA7)	aneurism	382313	2827	1982	16	14.7	1.09	0.57	1.16	0.68	1.82	1	

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
15q24_dup	ureter_bladder	332930	5	48530	3	0.7	2.95	0.020	6.00	1.37	22.99	182	0.35
15q24_dup	biliary	360684	6	20776	2	0.3	4.59	0.032	6.82	1.22	28.06	251	0.39
15q24_dup	heart_other	369638	7	11822	1	0.2	4.03	0.034	12.25	1.28	57.33	258	0.41
15q24_dup	hepatic	372706	7	8754	1	0.2	5.45	0.052	9.32	0.98	43.25	336	0.48
15q24_dup	died to 2016	370490	7	10970	1	0.2	4.35	0.061	8.45	0.88	39.62	367	0.52
15q24_dup	obesity	371622	7	9838	1	0.2	4.85	0.070	7.67	0.80	35.48	405	0.54
15q24_dup	thyroid	354318	6	27142	2	0.5	3.51	0.077	4.67	0.82	20.08	439	0.55
15q24_dup	venous_thromboembolic_disease	367487	7	13973	1	0.3	3.41	0.089	6.59	0.69	30.83	486	0.57
15q24_dup	allergy	322797	5	58663	3	0.9	2.44	0.094	3.43	0.79	13.02	504	0.59
15q24_dup	varicose_veins	369607	7	11853	1	0.2	4.02	0.096	6.29	0.66	29.37	509	0.59
15q24_dup	ear	367240	7	14220	1	0.3	3.35	0.096	6.27	0.66	29.15	510	0.59
15q24_dup	aneurism	379494	8	1966	0	0.0	0.00	0.11	33.02	0.25	274.22	571	0.63
15q24_dup	high_cholesterol	314878	6	66582	2	1.3	1.43	0.12	3.85	0.67	16.25	587	0.63
15q24_dup	prostate_hyperplasia	367013	8	14447	0	0.3	0.00	0.12	30.95	0.22	383.60	613	0.63
15q24_dup	arrhythmia	357301	7	24159	1	0.5	1.97	0.13	5.26	0.54	25.03	624	0.64
15q24_dup	irritable_bowel_syndrome	368456	7	13004	1	0.2	3.67	0.15	4.68	0.49	21.82	687	0.68
15q24_dup	respiratory	360709	7	20751	1	0.4	2.30	0.15	4.55	0.48	21.08	700	0.69
15q24_dup	sciatica	338347	6	43113	2	0.8	2.21	0.16	3.21	0.59	12.67	720	0.69
15q24_dup	migraine	367781	7	13679	1	0.3	3.49	0.16	4.38	0.46	20.42	733	0.70
15q24_dup	inflammatory_bowel_disease	362869	7	18591	1	0.4	2.56	0.19	3.98	0.42	18.32	803	0.73
15q24_dup	diverticular_disease_intestine	354326	7	27134	1	0.5	1.76	0.19	4.07	0.41	20.22	806	0.73
15q24_dup	paralytic_syndromes	379500	8	1960	0	0.0	0.00	0.21	12.36	0.09	102.91	879	0.75
15q24_dup	diabetes_insulin_dependent	378866	8	2594	0	0.1	0.00	0.23	10.41	0.08	85.25	944	0.78
15q24_dup	heartfailure	376385	8	5075	0	0.1	0.00	0.23	10.43	0.08	86.73	946	0.78
15q24_dup	nasal	378268	8	3192	0	0.1	0.00	0.24	9.84	0.08	79.73	975	0.78
15q24_dup	coagulation_defects	378919	8	2541	0	0.1	0.00	0.25	9.62	0.07	77.57	989	0.78
15q24_dup	endocrine_other	379100	8	2360	0	0.0	0.00	0.25	9.30	0.07	75.96	1008	0.78
15q24_dup	cerebrovascular_disease_not_stroke	377130	8	4330	0	0.1	0.00	0.26	9.22	0.07	79.60	1015	0.79
15q24_dup	gout	374370	8	7090	0	0.2	0.00	0.26	9.34	0.07	86.48	1017	0.79
15q24_dup	low_WBC_count	378495	8	2965	0	0.1	0.00	0.28	7.91	0.06	63.99	1088	0.80
15q24_dup	uterine_problem	339172	8	42288	0	1.0	0.00	0.31	0.29				

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2_del	diabetes_other	360794	76	20761	27	4.4	4.82	2.54E-11	6.07	3.77	9.50	2	3.98E-08
16p11.2_del	obesity	371699	84	9856	19	2.2	7.15	7.39E-10	6.81	4.01	11.01	3	7.71E-07
16p11.2_del	anaemia	362396	84	19159	19	4.4	3.68	2.15E-06	4.03	2.38	6.48	9	0.00075
16p11.2_del	hypertension	260873	52	120682	51	24.1	1.57	9.44E-06	2.55	1.69	3.83	12	0.0025
16p11.2_del	asthma	332199	71	49356	32	10.5	2.40	1.33E-05	2.71	1.77	4.08	14	0.0030
16p11.2_del	renal_failure	373625	93	7930	10	2.0	4.68	6.04E-05	5.11	2.49	9.45	16	0.012
16p11.2_del	osteoarthritis	312820	73	68735	30	16.0	1.62	0.00031	2.36	1.50	3.63	28	0.035
16p11.2_del	respiratory	360790	88	20765	15	5.1	2.68	0.0023	2.59	1.44	4.37	60	0.12
16p11.2_del	heartfailure	376474	97	5081	6	1.3	4.38	0.0045	4.16	1.64	8.84	76	0.19
16p11.2_del	died_to_2016	370576	93	10979	10	2.8	3.38	0.0050	2.88	1.42	5.27	79	0.20
16p11.2_del	high_cholesterol	314948	76	66607	27	16.1	1.50	0.0058	2.00	1.23	3.15	85	0.21
16p11.2_del	hernia	333645	81	47910	22	11.6	1.70	0.0084	1.99	1.20	3.16	105	0.25
16p11.2_del	atherosclerotic_vascular_disease	375800	97	5755	6	1.5	3.87	0.010	3.50	1.40	7.33	124	0.26
16p11.2_del	renal	374260	97	7295	6	1.9	3.05	0.015	3.16	1.28	6.49	160	0.30
16p11.2_del	gout	374459	97	7096	6	1.8	3.13	0.017	3.15	1.26	6.64	170	0.32
16p11.2_del	migraine	367869	95	13686	8	3.5	2.17	0.020	2.59	1.18	4.98	183	0.35
16p11.2_del	low_WBC_count	378587	100	2968	3	0.8	3.75	0.021	4.74	1.32	11.90	196	0.34
16p11.2_del	coagulation_defects	379011	100	2544	3	0.7	4.37	0.024	4.60	1.28	11.58	210	0.35
16p11.2_del	connective_tissue_disorder	372197	97	9358	6	2.4	2.38	0.029	2.79	1.13	5.76	240	0.38
16p11.2_del	ureter_bladder	333008	83	48547	20	12.1	1.53	0.034	1.76	1.05	2.81	257	0.41
16p11.2_del	psoriasis	375530	98	6025	5	1.6	3.08	0.039	2.86	1.06	6.16	285	0.43
16p11.2_del	arrhythmia	357386	92	24169	11	6.2	1.69	0.040	2.05	1.04	3.68	292	0.43
16p11.2_del	diabetes_insulin_dependent	378958	100	2597	3	0.7	4.28	0.045	3.75	1.04	9.48	310	0.45
16p11.2_del	cataract	359697	95	21858	8	5.8	1.36	0.064	2.14	0.95	4.23	377	0.53
16p11.2_del	ischaemic_heart_disease_not_MI	365224	95	16331	8	4.2	1.82	0.071	2.08	0.93	4.06	413	0.54
16p11.2_del	diverticular_disease_intestine	354420	101	27135	2	7.7	0.27	0.071	0.37	0.08	1.08	414	0.54
16p11.2_del	hepatic	372796	97	8759	6	2.3	2.54	0.077	2.24	0.91	4.61	435	0.55
16p11.2_del	allergy	322874	82	58681	21	14.9	1.33	0.081	1.56	0.94	2.46	459	0.55
16p11.2_del	digestive	299681	73	81874	30	19.9	1.36	0.099	1.45	0.93	2.20	526	0.59
16p11.2_del	COPD	367318	94	14237	9	3.6	2.34	0.12	1.83	0.85	3.52	583	0.63
16p11.2_del	uterine_problem												

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2_dup	irritable_bowel_syndrome	368567	118	13016	13	4.2	2.91	0.00036	3.33	1.80	5.67	30	0.037
16p11.2_dup	sciatica	338444	103	43139	28	13.1	1.89	0.0010	2.12	1.37	3.17	39	0.083
16p11.2_dup	venous_thromboembolic_disease	367600	120	13983	11	4.6	2.29	0.0093	2.49	1.28	4.37	119	0.24
16p11.2_dup	ureter_bladder	333030	105	48553	26	15.3	1.56	0.022	1.70	1.08	2.58	197	0.35
16p11.2_dup	renal_failure	373656	124	7927	7	2.6	2.57	0.025	2.65	1.15	5.23	216	0.36
16p11.2_dup	paralytic_syndromes	379620	128	1963	3	0.7	4.46	0.025	4.49	1.25	11.26	219	0.36
16p11.2_dup	heart_other	369754	123	11829	8	3.9	1.97	0.067	2.07	0.95	3.94	383	0.55
16p11.2_dup	hernia	333671	107	47912	24	15.4	1.46	0.068	1.55	0.97	2.38	393	0.54
16p11.2_dup	osteoporosis	369983	124	11600	7	3.9	1.76	0.083	2.10	0.90	4.21	463	0.56
16p11.2_dup	any_Ca	331877	121	49706	10	18.1	0.59	0.086	0.59	0.30	1.07	475	0.57
16p11.2_dup	aneurism	379615	129	1968	2	0.7	2.96	0.094	3.66	0.76	10.57	503	0.59
16p11.2_dup	stroke_derived2017	372759	125	8824	6	3.0	1.98	0.13	1.98	0.80	4.07	626	0.63
16p11.2_dup	psoriasis	375563	131	6020	0	2.1	0.00	0.16	0.22	0.00	1.53	729	0.70
16p11.2_dup	diverticular_disease_intestine	354437	118	27146	13	9.0	1.40	0.17	1.52	0.82	2.61	757	0.72
16p11.2_dup	anaemia	362433	121	19150	10	6.4	1.52	0.18	1.58	0.79	2.83	792	0.73
16p11.2_dup	obesity	371745	130	9838	1	3.4	0.30	0.20	0.40	0.05	1.46	826	0.74
16p11.2_dup	biliary	360799	121	20784	10	7.0	1.40	0.21	1.54	0.77	2.77	872	0.75
16p11.2_dup	ischaemic_heart_disease_not_MI	365257	128	16326	3	5.7	0.54	0.25	0.56	0.16	1.42	1003	0.78
16p11.2_dup	digestive	299705	97	81878	34	26.5	1.21	0.27	1.26	0.84	1.84	1043	0.80
16p11.2_dup	hepatic	372825	126	8758	5	3.0	1.66	0.27	1.69	0.63	3.62	1046	0.80
16p11.2_dup	renal	374290	127	7293	4	2.5	1.60	0.29	1.74	0.58	3.96	1126	0.81
16p11.2_dup	high_cholesterol	314984	112	66599	19	23.7	0.83	0.29	0.77	0.45	1.24	1139	0.81
16p11.2_dup	hypertension	260915	94	120668	37	43.5	0.89	0.33	0.82	0.55	1.21	1254	0.82
16p11.2_dup	prostate_hyperplasia	367133	128	14450	3	5.0	0.60	0.35	0.61	0.17	1.59	1305	0.83
16p11.2_dup	migraine	367899	125	13684	6	4.6	1.28	0.37	1.46	0.59	2.99	1385	0.85
16p11.2_dup	neuropathies	365673	124	15910	7	5.4	1.28	0.41	1.39	0.60	2.71	1486	0.86
16p11.2_dup	atherosclerotic_vascular_disease	375831	128	5752	3	2.0	1.52	0.42	1.60	0.44	4.04	1518	0.87
16p11.2_dup	ovarian_cysts	374192	128	7391	3	2.5	1.18	0.46	1.54	0.42	3.93	1644	0.88
16p11.2_dup	allergy	322907	115	58676	16	20.9	0.79	0.46	0.83	0.48	1.35	1651	0.88
16p11.2_dup	cerebrovascular_disease_not_stroke	377251	129	4332	2	1.5	1.34	0.47	1.65	0.34			

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2distal_del	diabetes_other	360757	39	20749	15	2.2	5.11	8.86E-08	6.95	3.67	12.57	6	4.63E-05
16p11.2distal_del	obesity	371660	45	9846	9	1.2	6.46	1.14E-05	7.06	3.27	13.73	13	0.0028
16p11.2distal_del	gout	374410	48	7096	6	0.9	5.98	0.00046	6.50	2.52	14.40	34	0.042
16p11.2distal_del	uterine_problem	339206	42	42300	12	5.2	2.00	0.0020	3.60	1.63	7.97	55	0.11
16p11.2distal_del	diabetes insulin dependent	378909	51	2597	3	0.3	8.17	0.0033	8.60	2.35	22.23	66	0.15
16p11.2distal_del	biliary	360725	47	20781	7	2.7	2.38	0.014	3.06	1.29	6.27	145	0.29
16p11.2distal_del	ovarian_cysts	374115	51	7391	3	1.0	2.87	0.050	3.77	1.00	10.35	329	0.47
16p11.2distal_del	heart_other	369681	50	11825	4	1.6	2.39	0.070	2.81	0.91	6.72	403	0.54
16p11.2distal_del	renal_failure	373583	51	7923	3	1.1	2.68	0.071	3.26	0.89	8.48	409	0.54
16p11.2distal_del	ureter_bladder	332968	43	48538	11	6.3	1.60	0.077	1.88	0.93	3.50	438	0.55
16p11.2distal_del	stroke_derived2017	372685	51	8821	3	1.2	2.40	0.10	2.86	0.78	7.41	532	0.60
16p11.2distal_del	hypertension	260853	32	120653	22	14.8	1.29	0.11	1.59	0.90	2.78	560	0.63
16p11.2distal_del	hepatic	372750	51	8756	3	1.2	2.42	0.13	2.63	0.72	6.77	623	0.64
16p11.2distal_del	paralytic_syndromes	379545	53	1961	1	0.3	3.60	0.14	4.66	0.53	17.22	654	0.65
16p11.2distal_del	high_cholesterol	314913	41	66593	13	8.7	1.38	0.15	1.64	0.83	3.05	683	0.68
16p11.2distal_del	COPD	367278	54	14228	0	2.1	0.00	0.16	0.22	0.00	1.54	727	0.69
16p11.2distal_del	coagulation_defects	378964	53	2542	1	0.4	2.78	0.17	3.96	0.45	14.57	754	0.72
16p11.2distal_del	ischaemic_heart_disease_not_MI	365179	50	16327	4	2.2	1.73	0.18	2.11	0.68	5.06	767	0.72
16p11.2distal_del	ear	367287	54	14219	0	2.1	0.00	0.18	0.23	0.00	1.62	781	0.73
16p11.2distal_del	cataract	359652	50	21854	4	3.0	1.29	0.20	2.03	0.65	4.92	833	0.75
16p11.2distal_del	varicose_veins	369651	51	11855	3	1.6	1.79	0.20	2.19	0.60	5.64	859	0.74
16p11.2distal_del	migraine	367828	54	13678	0	2.0	0.00	0.22	0.25	0.00	1.78	901	0.76
16p11.2distal_del	arrhythmia	357347	53	24159	1	3.6	0.29	0.24	0.42	0.05	1.57	947	0.78
16p11.2distal_del	died_to_2016	370534	51	10972	3	1.5	1.93	0.24	2.06	0.57	5.32	956	0.78
16p11.2distal_del	heart_valve	373128	52	8378	2	1.2	1.69	0.26	2.26	0.47	6.66	1039	0.79
16p11.2distal_del	diverticular_disease_intestine	354368	49	27138	5	3.8	1.30	0.28	1.70	0.62	3.80	1084	0.80
16p11.2distal_del	gastrointestinal_ulcer	371154	54	10352	0	1.5	0.00	0.32	0.31	0.00	2.19	1223	0.81
16p11.2distal_del	anaemia	362362	50	19144	4	2.6	1.48	0.36	1.63	0.53	3.85	1331	0.84
16p11.2distal_del	inflammatory_bowel_disease	362915	53	18591	1	2.7	0.38	0.39	0.53	0.06	1.94	1453	0.85
16p11.2distal_del	connective_tissue_disorder	372154	54	9352	0	1.4	0.00	0.45	0.40	0.00	2.77	1600	0.87

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2distal_dup	ischemic_heart_disease_not_MI	365245	116	16334	11	5.2	2.02	0.0085	2.55	1.30	4.56	107	0.25
16p11.2distal_dup	inflammatory_bowel_disease	362976	114	18603	13	5.8	2.10	0.0091	2.31	1.25	3.93	113	0.25
16p11.2distal_dup	heart_other	369749	118	11830	9	3.8	2.29	0.0091	2.77	1.32	5.15	115	0.25
16p11.2distal_dup	heart_valve	373196	120	8383	7	2.7	2.51	0.011	3.06	1.33	6.00	131	0.27
16p11.2distal_dup	endocrine_other	379216	124	2363	3	0.8	3.82	0.024	4.56	1.27	11.41	211	0.36
16p11.2distal_dup	diabetes_other	360843	125	20736	2	7.2	2.29	0.051	0.35	0.07	1.00	332	0.48
16p11.2distal_dup	stroke_derived2017	372755	121	8824	6	2.9	2.04	0.054	2.42	0.98	4.98	350	0.49
16p11.2distal_dup	gout	374484	122	7095	5	2.3	2.12	0.057	2.63	0.97	5.76	357	0.50
16p11.2distal_dup	uterine_problem	339283	119	42296	8	14.8	0.57	0.059	0.53	0.24	1.02	360	0.51
16p11.2distal_dup	connective_tissue_disorder	372227	127	9352	0	3.2	0.00	0.069	0.16	0.00	1.11	398	0.55
16p11.2distal_dup	arrhythmia	357409	115	24170	12	7.8	1.49	0.071	1.80	0.95	3.14	406	0.54
16p11.2distal_dup	any_Ca	331862	106	49717	21	15.9	1.27	0.071	1.58	0.96	2.48	408	0.54
16p11.2distal_dup	biliary	360794	116	20785	11	6.7	1.59	0.081	1.80	0.92	3.18	454	0.56
16p11.2distal_dup	hypertension	260902	81	120677	46	37.5	1.15	0.081	1.41	0.96	2.05	455	0.56
16p11.2distal_dup	gastrointestinal_ulcer	371221	121	10358	6	3.4	1.74	0.11	2.04	0.83	4.17	562	0.63
16p11.2distal_dup	diverticular_disease_intestine	354434	115	27145	12	8.8	1.33	0.13	1.64	0.86	2.86	622	0.64
16p11.2distal_dup	coagulation_defects	379036	125	2543	2	0.8	2.36	0.15	2.93	0.61	8.36	696	0.68
16p11.2distal_dup	ureter_bladder	333042	117	48537	10	17.1	0.62	0.16	0.65	0.32	1.17	723	0.69
16p11.2distal_dup	died_to_2016	370604	121	10975	6	3.6	1.64	0.18	1.80	0.73	3.67	784	0.73
16p11.2distal_dup	nasal	378385	125	3194	2	1.1	1.88	0.21	2.52	0.53	7.21	868	0.75
16p11.2distal_dup	obesity	371741	126	9838	1	3.3	0.31	0.21	0.41	0.05	1.49	873	0.75
16p11.2distal_dup	sciatica	338449	108	43130	19	13.8	1.32	0.28	1.33	0.78	2.12	1090	0.80
16p11.2distal_dup	ovarian_cysts	374187	123	7392	4	2.4	1.63	0.28	1.77	0.58	4.11	1101	0.80
16p11.2distal_dup	osteoporosis	369980	121	11599	6	3.8	1.55	0.30	1.64	0.60	3.62	1157	0.81
16p11.2distal_dup	aneurism	379612	126	1967	1	0.7	1.53	0.31	2.63	0.30	9.61	1197	0.81
16p11.2distal_dup	osteoarthritis	312848	101	68731	26	22.2	1.14	0.31	1.27	0.79	1.95	1204	0.81
16p11.2distal_dup	irritable_bowel_syndrome	368570	121	13009	6	4.3	1.39	0.35	1.49	0.61	3.04	1323	0.83
16p11.2distal_dup	ocular	359915	123	21664	4	7.4	0.55	0.35	0.66	0.22	1.51	1327	0.84
16p11.2distal_dup	psoriasis	375556	124	6023	3	2.0	1.50	0.36	1.71	0.48	4.26	1345	0.85
16p11.2distal_dup	paralytic_syndromes	379618	126	1961	1								

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p12.1_del	hypertension	260945	124	120742	111	57.4	1.49	8.64E-08	2.11	1.61	2.76	5	5.41E-05
16p12.1_del	obesity	371827	212	9860	23	5.6	3.80	1.11E-07	3.99	2.53	5.99	7	4.95E-05
16p12.1_del	renal_failure	373750	218	7937	17	4.6	3.48	7.23E-06	3.85	2.26	6.13	11	0.0021
16p12.1_del	diabetes_other	360926	208	20761	27	12.0	2.11	0.00021	2.34	1.53	3.46	24	0.028
16p12.1_del	heart_other	369848	217	11839	18	6.9	2.47	0.00029	2.77	1.65	4.36	27	0.034
16p12.1_del	ureter_bladder	333111	186	48576	49	27.1	1.64	0.00033	1.85	1.34	2.52	29	0.036
16p12.1_del	respiratory	360911	209	20776	26	12.0	2.03	0.00069	2.18	1.42	3.21	37	0.059
16p12.1_del	ischaemic_heart_disease_not_MI	365344	215	16343	20	9.6	1.99	0.0023	2.22	1.35	3.45	61	0.12
16p12.1_del	died_to_2016	370703	220	10984	15	6.5	2.22	0.0052	2.29	1.31	3.72	80	0.20
16p12.1_del	arrhythmia	357504	210	24183	25	14.2	1.68	0.0069	1.86	1.20	2.78	93	0.23
16p12.1_del	ovarian_cysts	374288	224	7399	11	4.4	2.42	0.0070	2.58	1.33	4.52	95	0.23
16p12.1_del	gastric_reflux	347359	202	34328	33	20.0	1.56	0.0098	1.67	1.14	2.38	123	0.25
16p12.1_del	COPD	367442	218	14245	17	8.5	1.94	0.010	2.04	1.20	3.27	127	0.26
16p12.1_del	uterine_problem	339361	197	42326	38	24.6	1.46	0.016	1.61	1.10	2.33	164	0.31
16p12.1_del	hernia	333758	194	47929	41	27.9	1.39	0.020	1.53	1.07	2.13	181	0.35
16p12.1_del	high_cholesterol	315054	182	66633	53	38.5	1.29	0.022	1.47	1.06	2.02	200	0.34
16p12.1_del	MI_derived2017	367812	220	13875	15	8.3	1.76	0.025	1.94	1.09	3.19	217	0.36
16p12.1_del	anaemia	362527	215	19160	20	11.4	1.70	0.027	1.74	1.07	2.68	226	0.37
16p12.1_del	osteoarthritis	312929	182	68758	53	40.0	1.25	0.066	1.35	0.98	1.84	382	0.55
16p12.1_del	venous_thromboembolic_disease	367701	221	13986	14	8.4	1.63	0.068	1.71	0.96	2.81	388	0.55
16p12.1_del	atherosclerotic_vascular_disease	375931	228	5756	7	3.5	1.98	0.070	2.13	0.93	4.13	402	0.55
16p12.1_del	ear	367454	221	14233	14	8.6	1.60	0.075	1.69	0.95	2.77	423	0.55
16p12.1_del	diabetes_insulin_dependent	379089	231	2598	4	1.6	2.50	0.075	2.68	0.89	6.06	424	0.55
16p12.1_del	inflammatory_bowel_disease	363091	229	18596	6	11.7	0.52	0.081	0.53	0.22	1.07	456	0.56
16p12.1_del	heartfailure	376606	229	5081	6	3.1	1.92	0.092	2.13	0.87	4.33	498	0.58
16p12.1_del	digestive	299782	174	81905	61	47.5	1.21	0.10	1.28	0.95	1.71	536	0.61
16p12.1_del	cerebrovascular_disease_not_stroke	377352	230	4335	5	2.6	1.87	0.11	2.16	0.81	4.59	567	0.63
16p12.1_del	paralytic_syndromes	379724	232	1963	3	1.2	2.48	0.11	2.69	0.75	6.66	569	0.63
16p12.1_del	gastrointestinal_ulcer	371325	225	10362	10	6.3	1.57	0.14	1.67	0.84	2.95	656	0.65
16p12.1_del	diverticular_disease_intestine	354532	213	27155	22	16.3	1.32	0.14	1.42	0.89	2.16	658	0.65
16p12.1_del	biliary	360895	217	20792	18	12.5	1.41	0.14	1.46	0.88	2.29	667	0.66
16p12.1_del	gout	374590	228	7097	7	4.3	1.60	0.15	1.82	0.79	3.56	680	0.67
16p12.1_del	thyroid	354525	213	27162	22	16.3	1.32	0.17	1.38	0.86	2.10	758	0.72
16p12.1_del	osteoporosis	370084	225	11603	10	7.1	1.40	0.22	1.52	0.76	2.72	909	0.77
16p12.1_del	prostate_hyperplasia	367229	224	14458	11	8.8	1.24	0.24	1.49	0.75	2.70	971	0.78
16p12.1_del	sciatica	338545	204	43142	31	26.0	1.17	0.37	1.19	0.80	1.71	1374	0.85
16p12.1_del	migraine	368003	229	13684	6	8.5	0.71	0.43	0.74	0.30	1.49	1558	0.87
16p12.1_del	hepatic	372927	228	8760	7	5.4	1.30	0.44	1.35	0.59	2.60	1587	0.87
16p12.1_del	any_Ca	331958	202	49729	33	30.3	1.08	0.47	1.15	0.78	1.64	1683	0.88
16p12.1_del	psoriasis	375665	233	6022	2	3.7	0.54	0.48	0.66	0.14	1.85	1687	0.88
16p12.1_del	coagulation_defects	379144	233	2543	2	1.6	1.28	0.52	1.55	0.33	4.40	1807	0.90
16p12.1_del	cataract	359822	220	21865	15	13.4	1.11	0.55	1.18	0.67	1.93	1893	0.92
16p12.1_del	asthma	332330	202	49357	33	30.0	1.09	0.63	1.10	0.75	1.56	2095	0.94
16p12.1_del	low_WBC_count	378720	233	2967	2	1.8	1.09	0.64	1.36	0.29	3.85	2130	0.95
16p12.1_del	congenital	376658	233	5029	2	3.1	0.65	0.67	0.77	0.16	2.18	2217	0.95
16p12.1_del	renal	374393	230	7294	5	4.5	1.11	0.69	1.20	0.45	2.53	2258	0.95
16p12.1_del	stroke_derived2017	372863	229	8824	6	5.4	1.10	0.69	1.17	0.48	2.37	2279	0.95
16p12.1_del	ocular	360013	221	21674	14	13.3	1.05	0.74	1.10	0.62	1.80	2396	0.96
16p12.1_del	irritable_bowel_syndrome	368677	228	13010	7	8.0	0.87	0.77	0.90	0.40	1.74	2509	0.97
16p12.1_del	nasal	378494	234	3193	1	2.0	0.51	0.78	0.80	0.09	2.87	2527	0.97
16p12.1_del	aneurism	379720	234	1967	1	1.2	0.83	0.79	1.25	0.14	4.52	2563	0.97
16p12.1_del	heart_valve	373307	231	8380	4	5.2	0.78	0.79	0.88	0.29	2.00	2565	0.97
16p12.1_del	connective_tissue_disorder	372329	229	9358	6	5.8	1.04	0.79	1.11	0.46	2.24	2569	0.97
16p12.1_del	glaucoma	375156	232	6531	3	4.0	0.75	0.81	0.88	0.25	2.19	2620	0.97
16p12.1_del	neuropathies	365774	225	15913	10	9.8	1.02	0.90	1.04	0.52	1.83	2885	0.98
16p12.1_del	allergy	322992	200	58695	35	36.3	0.97	0.97	0.99	0.68	1.40	3051	0.99
16p12.1_del	varicose_veins	369828	228	11859	7	7.3	0.96	0.99	1.00	0.44	1.93	3111	1.00
16p12.1_del	endocrine_other	379326	234	2361	1	1.5	0.69	0.99	1.01	0.12	3.61	3114	1.00



CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p12.1_dup	high_cholesterol	315044	172	66600	20	36.4	0.60	0.0083	0.55	0.33	0.86	104	0.25
16p12.1_dup	osteoarthritis	312893	146	68751	46	32.1	1.33	0.025	1.50	1.06	2.10	213	0.36
16p12.1_dup	asthma	332285	157	49359	35	23.3	1.41	0.037	1.50	1.03	2.14	272	0.42
16p12.1_dup	any_Ca	331933	177	49711	15	26.5	0.60	0.038	0.60	0.34	0.97	280	0.43
16p12.1_dup	heart_other	369813	182	11831	10	5.8	1.68	0.073	1.87	0.94	3.35	422	0.54
16p12.1_dup	psoriasis	375618	186	6026	6	3.0	1.98	0.080	2.20	0.90	4.42	451	0.55
16p12.1_dup	cataract	359778	176	21866	16	10.7	1.45	0.090	1.61	0.92	2.64	488	0.58
16p12.1_dup	ocular	359968	176	21676	16	10.6	1.47	0.091	1.60	0.92	2.57	492	0.58
16p12.1_dup	varicose_veins	369782	182	11862	10	5.8	1.68	0.091	1.79	0.90	3.18	494	0.58
16p12.1_dup	irritable_bowel_syndrome	368630	181	13014	11	6.4	1.68	0.093	1.74	0.90	3.03	499	0.59
16p12.1_dup	migraine	367963	189	13681	3	7.0	0.44	0.12	0.47	0.13	1.17	576	0.63
16p12.1_dup	gout	374548	186	7096	6	3.5	1.68	0.12	2.00	0.81	4.11	611	0.63
16p12.1_dup	nasal	378449	189	3195	3	1.6	1.87	0.18	2.27	0.63	5.64	778	0.72
16p12.1_dup	uterine_problem	339339	175	42305	17	21.8	0.80	0.18	0.72	0.42	1.16	788	0.73
16p12.1_dup	died_to_2016	370667	184	10977	8	5.4	1.45	0.22	1.59	0.73	2.98	897	0.76
16p12.1_dup	CPD	367406	182	14238	10	7.1	1.40	0.25	1.49	0.74	2.68	994	0.78
16p12.1_dup	glaucoma	375111	187	6533	5	3.3	1.52	0.25	1.72	0.64	3.66	1009	0.78
16p12.1_dup	neuropathies	365730	181	15914	11	7.9	1.37	0.27	1.43	0.74	2.48	1052	0.80
16p12.1_dup	ear	367421	188	14223	4	7.3	0.56	0.28	0.62	0.21	1.41	1098	0.80
16p12.1_dup	prostate_hyperplasia	367193	188	14451	4	7.4	0.55	0.28	0.61	0.20	1.44	1103	0.80
16p12.1_dup	ureter_bladder	333098	173	48546	19	25.2	0.78	0.30	0.78	0.47	1.22	1143	0.81
16p12.1_dup	endocrine_other	379282	190	2362	2	1.2	1.68	0.31	2.07	0.43	5.88	1181	0.81
16p12.1_dup	hepatic	372889	190	8755	2	4.5	0.45	0.31	0.56	0.12	1.58	1189	0.82
16p12.1_dup	sciatica	338515	174	43129	18	22.2	0.83	0.31	0.78	0.46	1.24	1209	0.81
16p12.1_dup	hypertension	260960	139	120684	53	64.3	0.87	0.32	0.85	0.60	1.17	1219	0.82
16p12.1_dup	respiratory	360887	185	20757	7	10.6	0.67	0.33	0.71	0.31	1.38	1269	0.82
16p12.1_dup	coagulation_defects	379101	190	2543	2	1.3	1.56	0.34	1.97	0.41	5.57	1288	0.82
16p12.1_dup	gastric_reflux	347336	179	34308	13	17.7	0.75	0.34	0.77	0.42	1.29	1300	0.83
16p12.1_dup	allergy	322950	158	58694	34	28.7	1.15	0.35	1.20	0.81	1.71	1321	0.83
16p12.1_dup	diabetes_insulin_dependent	379048	190	2596	2	1.3	1.53	0.36	1.92	0.40	5.45	1329	0.84
16p12.1_dup	heart_valve	373266	190	8378	2	4.3	0.47	0.37	0.59	0.12	1.68	1373	0.85
16p12.1_dup	cerebrovascular_disease_not_stroke	377311	189	4333	3	2.2	1.38	0.38	1.66	0.46	4.15	1419	0.85
16p12.1_dup	renal_failure	373722	190	7922	2	4.0	0.50	0.42	0.62	0.13	1.78	1532	0.87
16p12.1_dup	digestive	299764	156	81880	36	42.6	0.87	0.43	0.87	0.59	1.23	1549	0.87
16p12.1_dup	obesity	371804	189	9840	3	5.0	0.61	0.44	0.68	0.19	1.68	1577	0.87
16p12.1_dup	low_WBC_count	378677	190	2967	2	1.5	1.34	0.45	1.68	0.35	4.76	1622	0.88
16p12.1_dup	gastrointestinal_ulcer	371289	189	10355	3	5.3	0.58	0.46	0.68	0.19	1.70	1626	0.88
16p12.1_dup	heartfailure	376568	191	5076	1	2.6	0.39	0.49	0.59	0.07	2.14	1724	0.88
16p12.1_dup	inflammatory_bowel_disease	363047	185	18597	7	9.5	0.75	0.49	0.78	0.34	1.51	1745	0.89
16p12.1_dup	osteoporosis	370044	185	11600	7	5.8	1.20	0.51	1.30	0.56	2.55	1776	0.89
16p12.1_dup	hernia	333730	166	47914	26	23.8	1.08	0.51	1.15	0.74	1.72	1780	0.89
16p12.1_dup	connective_tissue_disorder	372289	189	9355	3	4.7	0.64	0.53	0.72	0.20	1.79	1832	0.90
16p12.1_dup	ischaemic_heart_disease_not_MI	365315	186	16329	6	8.3	0.73	0.55	0.79	0.32	1.61	1891	0.92
16p12.1_dup	biliary	360858	180	20786	12	10.4	1.15	0.58	1.18	0.63	2.02	1952	0.93
16p12.1_dup	congenital	376614	189	5030	3	2.5	1.19	0.59	1.36	0.38	3.37	1981	0.93
16p12.1_dup	venous_thromboembolic_disease	367664	184	13980	8	7.0	1.14	0.60	1.21	0.56	2.27	2005	0.93
16p12.1_dup	aneurism	379678	192	1966	0	1.0	0.00	0.61	0.52	0.00	3.58	2029	0.94
16p12.1_dup	paralytic_syndromes	379683	191	1961	1	1.0	1.01	0.64	1.51	0.17	5.43	2115	0.94
16p12.1_dup	stroke_derived2017	372823	189	8821	3	4.5	0.68	0.65	0.79	0.22	1.96	2142	0.95
16p12.1_dup	atherosclerotic_vascular_disease	375892	189	5752	3	2.9	1.04	0.72	1.22	0.34	3.05	2335	0.96
16p12.1_dup	ovarian_cysts	374253	189	7391	3	3.7	0.81	0.76	0.85	0.24	2.12	2456	0.96
16p12.1_dup	MI_derived2017	367777	185	13867	7	7.0	1.00	0.76	1.12	0.49	2.21	2470	0.97
16p12.1_dup	anaemia	362494	182	19150	10	9.6	1.04	0.84	1.07	0.54	1.90	2687	0.97
16p12.1_dup	thyroid	354491	179	27153	13	13.7	0.95	0.86	0.95	0.52	1.60	2763	0.97
16p12.1_dup	arrhythmia	357475	181	24169	11	12.2	0.90	0.91	0.96	0.50	1.68	2892	0.98
16p12.1_dup	renal	374352	189	7292	3	3.7	0.82	0.93	0.95	0.27	2.36	2948	0.98
16p12.1_dup	diverticular_disease_intestine	354498	179	27146	13	13.7	0.95	0.97	1.01	0.55	1.70	3060	0.99
16p12.1_dup	diabetes_other	360900	182	20744	10	10.5	0.96	0.97	1.01	0.51	1.81	3075	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p13.11_del	obesity	371729	114	9847	10	3.0	3.13	0.0013	3.41	1.70	6.15	42	0.096
16p13.11_del	stroke_derived2017	372750	116	8826	8	2.7	2.79	0.0049	3.28	1.49	6.29	78	0.20
16p13.11_del	low_WBC_count	378607	120	2969	4	0.9	4.15	0.0095	4.69	1.56	10.72	121	0.25
16p13.11_del	asthma	332226	98	49350	26	14.6	1.62	0.013	1.78	1.14	2.69	144	0.28
16p13.11_del	anaemia	362423	111	19153	13	5.9	2.09	0.014	2.21	1.19	3.75	146	0.29
16p13.11_del	digestive	299696	88	81880	36	24.0	1.35	0.035	1.55	1.03	2.26	262	0.41
16p13.11_del	osteoporosis	369975	116	11601	8	3.6	2.12	0.044	2.26	1.02	4.38	307	0.45
16p13.11_del	hepatic	372817	118	8759	6	2.8	2.11	0.061	2.35	0.95	4.78	369	0.52
16p13.11_del	ureter_bladder	333027	102	48549	22	14.9	1.39	0.068	1.57	0.97	2.44	391	0.55
16p13.11_del	paralytic_syndromes	379614	122	1962	2	0.6	3.14	0.079	3.95	0.82	11.30	447	0.55
16p13.11_del	ovarian_cysts	374188	124	7388	0	2.4	0.00	0.082	0.17	0.00	1.17	460	0.56
16p13.11_del	sciatica	338445	104	43131	20	13.3	1.43	0.097	1.53	0.92	2.40	516	0.59
16p13.11_del	thyroid	354422	110	27154	14	8.4	1.59	0.12	1.60	0.88	2.71	594	0.63
16p13.11_del	cerebrovascular_disease_not_stroke	377243	121	4333	3	1.4	2.13	0.12	2.63	0.73	6.65	614	0.63
16p13.11_del	glaucoma	375044	120	6532	4	2.1	1.88	0.13	2.27	0.75	5.23	648	0.64
16p13.11_del	respiratory	360816	114	20760	10	6.6	1.48	0.15	1.66	0.82	2.99	679	0.67
16p13.11_del	heart_other	369749	118	11827	6	3.8	1.56	0.17	1.85	0.75	3.81	736	0.70
16p13.11_del	gout	374486	124	7090	0	2.3	0.00	0.19	0.24	0.00	1.67	820	0.74
16p13.11_del	gastric_reflux	347266	109	34310	15	10.8	1.35	0.20	1.45	0.81	2.39	828	0.74
16p13.11_del	venous_thromboembolic_disease	367597	117	13979	7	4.4	1.54	0.20	1.69	0.73	3.31	836	0.74
16p13.11_del	nasal	378382	122	3194	2	1.0	1.93	0.21	2.53	0.53	7.23	867	0.75
16p13.11_del	biliary	360792	114	20784	10	6.6	1.48	0.21	1.53	0.76	2.76	884	0.76
16p13.11_del	psoriasis	375556	124	6020	0	2.0	0.00	0.21	0.25	0.00	1.74	885	0.76
16p13.11_del	congenital	376546	121	5030	3	1.6	1.84	0.22	2.12	0.59	5.29	891	0.76
16p13.11_del	connective_tissue_disorder	372219	119	9357	5	3.0	1.64	0.23	1.78	0.66	3.82	924	0.77
16p13.11_del	renal_failure	373652	120	7924	4	2.5	1.55	0.24	1.86	0.61	4.30	967	0.78
16p13.11_del	atherosclerotic_vascular_disease	375827	124	5749	0	1.9	0.00	0.24	0.27	0.00	1.86	972	0.78
16p13.11_del	ischaemic_heart_disease_not_MI	365246	117	16330	7	5.2	1.32	0.29	1.54	0.66	3.07	1122	0.81
16p13.11_del	heart_valve	373196	120	8380	4	2.7	1.47	0.30	1.72	0.57	3.94	1155	0.81
16p13.11_del	heartfailure	376501	124	5075	0	1.7	0.00	0.32	0.32	0.00	2.19	1224	0.81
16p13.11_del	cataract	359717	115	21859	9	7.0	1.27	0.32	1.44	0.68	2.70	1225	0.81
16p13.11_del	diverticular_disease_intestine	354432	113	27144	11	8.7	1.25	0.33	1.37	0.70	2.44	1262	0.82
16p13.11_del	prostate_hyperplasia	367124	119	14452	5	4.7	1.06	0.42	1.48	0.53	3.46	1527	0.87
16p13.11_del	arrhythmia	357413	119	24163	5	8.0	0.64	0.43	0.72	0.27	1.55	1542	0.87
16p13.11_del	diabetes_other	360834	116	20742	8	6.7	1.19	0.44	1.34	0.61	2.57	1564	0.87
16p13.11_del	uterine_problem	339275	111	42301	13	13.8	0.95	0.45	0.80	0.43	1.39	1603	0.87
16p13.11_del	migraine	367892	118	13684	6	4.4	1.35	0.47	1.36	0.55	2.77	1667	0.88
16p13.11_del	osteoarthritis	312846	99	68730	25	21.7	1.12	0.48	1.18	0.74	1.82	1686	0.88
16p13.11_del	renal	374284	121	7292	3	2.4	1.27	0.50	1.48	0.41	3.69	1751	0.89
16p13.11_del	diabetes_insulin_dependent	378981	123	2595	1	0.8	1.19	0.51	1.80	0.21	6.52	1799	0.89
16p13.11_del	any_Ca	331867	111	49709	13	16.6	0.80	0.54	0.84	0.45	1.44	1865	0.91
16p13.11_del	ocular	359908	116	21668	8	7.0	1.14	0.57	1.23	0.57	2.34	1941	0.92
16p13.11_del	died_to_2016	370603	120	10973	4	3.6	1.12	0.57	1.33	0.44	3.04	1945	0.92
16p13.11_del	ear	367354	121	14222	3	4.7	0.65	0.58	0.75	0.21	1.87	1947	0.93
16p13.11_del	high_cholesterol	314974	102	66602	22	21.6	1.02	0.64	1.12	0.68	1.78	2124	0.95
16p13.11_del	irritable_bowel_syndrome	368568	119	13008	5	4.2	1.18	0.68	1.20	0.45	2.58	2232	0.95
16p13.11_del	allergy	322895	103	58681	21	18.7	1.10	0.68	1.10	0.68	1.72	2233	0.95
16p13.11_del	neuropathies	365669	120	15907	4	5.2	0.77	0.69	0.83	0.27	1.89	2260	0.95
16p13.11_del	coagulation_defects	379035	124	2541	0	0.8	0.00	0.69	0.60	0.00	4.07	2267	0.95
16p13.11_del	endocrine_other	379216	124	2360	0	0.8	0.00	0.72	0.62	0.00	4.26	2333	0.96
16p13.11_del	hypertension	260906	85	120670	39	39.3	0.99	0.74	1.07	0.71	1.58	2411	0.96
16p13.11_del	inflammatory_bowel_disease	362981	119	18595	5	6.1	0.83	0.77	0.88	0.33	1.89	2497	0.97
16p13.11_del	varicose_veins	369721	121	11855	3	3.9	0.78	0.79	0.87	0.24	2.17	2561	0.97
16p13.11_del	MI_derived2017	367712	120	13864	4	4.5	0.89	0.84	1.11	0.36	2.61	2686	0.97
16p13.11_del	gastrointestinal_ulcer	371221	121	10355	3	3.4	0.89	0.90	1.07	0.30	2.69	2878	0.98
16p13.11_del	hernia	333674	110	47902	14	15.8	0.90	0.92	0.97	0.53	1.64	2927	0.98
16p13.11_del	aneurism	379610	124	1966	0	0.6	0.00	0.92	0.87	0.01	6.02	2929	0.98
16p13.11_del	COPD	367344	120	14232	4	4.6	0.86	0.97	0.98	0.32	2.28	3052	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p13.11_dup	hypertension	261304	483	120931	300	223.5	1.21	2.05E-05	1.39	1.20	1.62	15	0.0043
16p13.11_dup	died_to_2016	371226	743	11009	40	22.0	1.78	0.00097	1.79	1.28	2.43	38	0.080
16p13.11_dup	diabetes_other	361437	719	20798	64	41.4	1.50	0.0018	1.55	1.18	1.99	52	0.11
16p13.11_dup	ureter_bladder	333580	655	48655	128	95.5	1.29	0.0023	1.36	1.12	1.64	59	0.12
16p13.11_dup	prostate_hyperplasia	367748	743	14487	40	29.3	1.35	0.011	1.58	1.11	2.19	130	0.27
16p13.11_dup	endocrine_other	379866	774	2369	9	4.8	1.86	0.064	1.96	0.96	3.51	375	0.53
16p13.11_dup	diverticular_disease_intestine	355035	716	27200	67	54.9	1.20	0.065	1.28	0.98	1.64	378	0.54
16p13.11_dup	gout	375124	762	7111	21	14.4	1.44	0.082	1.51	0.95	2.27	461	0.56
16p13.11_dup	digestive	300242	634	81993	149	173.1	0.89	0.094	0.86	0.72	1.03	505	0.58
16p13.11_dup	renal_failure	374293	761	7942	22	16.1	1.35	0.15	1.39	0.88	2.07	678	0.67
16p13.11_dup	glaucoma	375699	775	6536	8	13.5	0.60	0.19	0.66	0.31	1.21	809	0.74
16p13.11_dup	migraine	368536	762	13699	21	28.3	0.75	0.19	0.76	0.48	1.14	821	0.74
16p13.11_dup	aneurism	380263	777	1972	6	4.0	1.49	0.24	1.66	0.68	3.31	949	0.78
16p13.11_dup	any_Ca	332430	674	49805	109	101.0	1.07	0.26	1.13	0.91	1.38	1029	0.79
16p13.11_dup	heart_other	370385	754	11850	29	24.1	1.20	0.27	1.24	0.84	1.76	1074	0.80
16p13.11_dup	heart_valve	373838	762	8397	21	17.1	1.22	0.28	1.28	0.81	1.92	1092	0.80
16p13.11_dup	COPD	367971	747	14264	36	29.0	1.23	0.29	1.21	0.85	1.67	1120	0.80
16p13.11_dup	inflammatory_bowel_disease	363613	751	18622	32	38.5	0.84	0.30	0.83	0.57	1.16	1142	0.81
16p13.11_dup	anaemia	363062	750	19173	33	39.6	0.84	0.30	0.84	0.58	1.16	1145	0.81
16p13.11_dup	arrhythmia	358022	728	24213	55	49.2	1.11	0.31	1.16	0.87	1.51	1208	0.81
16p13.11_dup	biliary	361425	747	20810	36	43.0	0.84	0.33	0.85	0.60	1.17	1261	0.82
16p13.11_dup	asthma	332799	671	49436	112	99.7	1.11	0.34	1.10	0.90	1.34	1293	0.83
16p13.11_dup	obesity	372733	758	9862	25	20.1	1.24	0.35	1.22	0.80	1.77	1311	0.83
16p13.11_dup	hernia	334241	677	47994	106	97.2	1.08	0.36	1.10	0.89	1.35	1340	0.84
16p13.11_dup	heartfailure	377147	770	5088	13	10.4	1.25	0.37	1.30	0.72	2.14	1362	0.85
16p13.11_dup	allergy	323463	671	58772	112	121.9	0.93	0.43	0.92	0.75	1.12	1540	0.87
16p13.11_dup	thyroid	355035	723	27200	60	55.4	1.08	0.44	1.11	0.84	1.44	1571	0.87
16p13.11_dup	hepatic	373467	768	8768	15	18.0	0.83	0.47	0.83	0.48	1.33	1659	0.88
16p13.11_dup	ovarian_cysts	374835	771	7400	12	15.2	0.79	0.50	0.83	0.45	1.39	1748	0.89
16p13.11_dup	sciatica	339041	700	43194	83	89.2	0.94	0.50	0.93	0.73	1.15	1759	0.89
16p13.11_dup	congenital	377200	775	5035	8	10.3	0.78	0.52	0.81	0.38	1.48	1815	0.90
16p13.11_dup	respiratory	361438	736	20797	47	42.3	1.10	0.52	1.10	0.81	1.46	1825	0.90
16p13.11_dup	connective_tissue_disorder	372862	762	9373	21	19.2	1.09	0.61	1.12	0.71	1.68	2022	0.94
16p13.11_dup	diabetes_insulin_dependent	379637	779	2598	4	5.3	0.75	0.62	0.80	0.27	1.79	2069	0.94
16p13.11_dup	gastric_reflux	347874	717	34361	66	70.8	0.94	0.62	0.94	0.72	1.20	2071	0.94
16p13.11_dup	ischaemic_heart_disease_not_MI	365877	748	16358	35	33.4	1.04	0.64	1.09	0.76	1.50	2117	0.94
16p13.11_dup	neuropathies	366302	753	15933	30	32.8	0.92	0.64	0.92	0.63	1.30	2129	0.95
16p13.11_dup	MI_derived2017	368345	753	13890	30	28.4	1.05	0.69	1.08	0.73	1.53	2276	0.95
16p13.11_dup	cerebrovascular_disease_not_stroke	377898	776	4337	7	8.9	0.79	0.71	0.87	0.39	1.67	2305	0.96
16p13.11_dup	coagulation_defects	379690	779	2545	4	5.2	0.77	0.72	0.85	0.28	1.90	2334	0.96
16p13.11_dup	renal	374930	767	7305	16	14.9	1.07	0.72	1.09	0.64	1.72	2345	0.96
16p13.11_dup	ocular	360534	742	21701	41	44.7	0.92	0.73	0.95	0.68	1.28	2373	0.96
16p13.11_dup	uterine_problem	339865	701	42370	82	87.4	0.94	0.73	0.96	0.75	1.21	2387	0.96
16p13.11_dup	irritable_bowel_syndrome	369204	755	13031	28	26.6	1.05	0.74	1.07	0.72	1.52	2395	0.96
16p13.11_dup	venous_thromboembolic_disease	368233	753	14002	30	28.6	1.05	0.75	1.06	0.72	1.50	2427	0.97
16p13.11_dup	stroke_derived2017	373398	764	8837	19	18.1	1.05	0.79	1.07	0.65	1.63	2543	0.97
16p13.11_dup	cataract	360341	739	21894	44	44.9	0.98	0.80	1.04	0.75	1.40	2578	0.97
16p13.11_dup	gastrointestinal_ulcer	371861	761	10374	22	21.2	1.04	0.82	1.05	0.67	1.56	2644	0.97
16p13.11_dup	low_WBC_count	379264	777	2971	6	6.1	0.99	0.85	1.08	0.45	2.15	2729	0.97
16p13.11_dup	paralytic_syndromes	380271	779	1964	4	4.0	0.99	0.89	1.07	0.36	2.40	2825	0.98
16p13.11_dup	osteoarthritis	313390	643	68845	140	141.3	0.99	0.90	1.01	0.84	1.22	2877	0.98
16p13.11_dup	atherosclerotic_vascular_disease	376474	771	5761	12	11.8	1.02	0.91	1.03	0.56	1.73	2891	0.98
16p13.11_dup	varicose_veins	370359	759	11876	24	24.3	0.99	0.93	1.02	0.66	1.49	2949	0.98
16p13.11_dup	osteoporosis	370619	760	11616	23	23.8	0.97	0.94	1.02	0.65	1.51	2985	0.99
16p13.11_dup	high_cholesterol	315520	648	66715	135	137.0	0.99	0.95	1.01	0.83	1.22	3012	0.99
16p13.11_dup	ear	367987	754	14248	29	29.2	0.99	0.96	1.01	0.68	1.43	3028	0.99
16p13.11_dup	psoriasis	376203	771	6032	12	12.4	0.97	0.97	0.99	0.53	1.65	3053	0.99
16p13.11_dup	nasal	379037	777	3198	6	6.6	0.92	0.98	1.01	0.42	2.01	3086	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
17p12_CMT1A_dup	neuropathies	365609	60	15959	56	2.6	11.58	3.94E-124	21.75	15.02	31.46	1	1.23E-120
17p12_CMT1A_dup	anaemia	362411	99	19157	17	5.2	2.92	6.70E-05	3.30	1.91	5.36	18	0.012
17p12_CMT1A_dup	stroke_derived2017	372741	107	8827	9	2.5	3.36	0.0013	3.70	1.76	6.92	43	0.094
17p12_CMT1A_dup	digestive	299685	77	81883	39	21.0	1.57	0.0017	1.91	1.29	2.81	48	0.11
17p12_CMT1A_dup	osteoarthritis	312830	83	68738	33	18.2	1.58	0.0029	1.94	1.26	2.93	64	0.14
17p12_CMT1A_dup	MI_derived2017	367697	105	13871	11	4.0	2.61	0.0032	2.96	1.48	5.40	65	0.15
17p12_CMT1A_dup	heartfailure	376487	110	5081	6	1.5	3.89	0.0034	4.32	1.73	9.03	68	0.16
17p12_CMT1A_dup	ureter_bladder	333016	91	48552	25	13.3	1.69	0.0054	1.96	1.23	3.01	81	0.21
17p12_CMT1A_dup	sciatica	338451	110	43117	6	14.0	0.46	0.028	0.45	0.18	0.93	234	0.37
17p12_CMT1A_dup	arrhythmia	357397	103	24171	13	7.0	1.77	0.030	2.00	1.07	3.45	244	0.39
17p12_CMT1A_dup	renal_failure	373642	110	7926	6	2.3	2.49	0.035	2.69	1.08	5.55	263	0.41
17p12_CMT1A_dup	prostate_hyperplasia	367113	108	14455	8	4.3	1.82	0.046	2.34	1.02	4.82	315	0.45
17p12_CMT1A_dup	COPD	367331	107	14237	9	4.1	2.08	0.053	2.11	0.99	3.99	346	0.48
17p12_CMT1A_dup	connective_tissue_disorder	372210	110	9358	6	2.8	2.11	0.059	2.37	0.96	4.87	362	0.51
17p12_CMT1A_dup	gastric_reflux	347257	100	34311	16	9.9	1.53	0.064	1.69	0.97	2.78	376	0.53
17p12_CMT1A_dup	paralytic_syndromes	379606	114	1962	2	0.6	3.36	0.081	3.89	0.81	11.18	458	0.56
17p12_CMT1A_dup	gastrointestinal_ulcer	371210	110	10358	6	3.1	1.91	0.090	2.16	0.87	4.43	490	0.58
17p12_CMT1A_dup	endocrine_other	379206	114	2362	2	0.7	2.79	0.11	3.46	0.72	9.91	538	0.61
17p12_CMT1A_dup	hernia	333660	96	47908	20	13.8	1.37	0.12	1.50	0.90	2.39	582	0.63
17p12_CMT1A_dup	gout	374474	112	7094	4	2.1	1.86	0.17	2.11	0.69	4.96	745	0.71
17p12_CMT1A_dup	congenital	376538	113	5030	3	1.5	1.96	0.19	2.22	0.62	5.56	812	0.74
17p12_CMT1A_dup	any_Ca	331854	98	49714	18	14.7	1.19	0.27	1.35	0.79	2.18	1045	0.80
17p12_CMT1A_dup	allergy	322886	94	58682	22	17.1	1.23	0.27	1.31	0.80	2.03	1049	0.80
17p12_CMT1A_dup	osteoporosis	369970	111	11598	5	3.5	1.42	0.27	1.70	0.62	3.74	1056	0.80
17p12_CMT1A_dup	inflammatory_bowel_disease	362970	108	18598	8	5.5	1.42	0.28	1.50	0.69	2.84	1107	0.80
17p12_CMT1A_dup	venous_thromboembolic_disease	367590	110	13978	6	4.2	1.41	0.33	1.52	0.62	3.10	1263	0.82
17p12_CMT1A_dup	ear	367343	110	14225	6	4.3	1.39	0.33	1.52	0.62	3.10	1264	0.82
17p12_CMT1A_dup	varicose_veins	369711	111	11857	5	3.6	1.39	0.33	1.57	0.59	3.37	1268	0.82
17p12_CMT1A_dup	cerebrovascular_disease_not_stroke	377236	114	4332	2	1.3	1.52	0.34	1.98	0.41	5.71	1274	0.83
17p12_CMT1A_dup	aneurism	379601	115	1967	1	0.6	1.67	0.35	2.43	0.28	8.94	1308	0.83
17p12_CMT1A_dup	ocular	359904	112	21664	4	6.7	0.61	0.38	0.67	0.22	1.54	1400	0.85
17p12_CMT1A_dup	asthma	332232	104	49336	12	15.4	0.80	0.39	0.78	0.41	1.34	1421	0.85
17p12_CMT1A_dup	respiratory	360810	108	20758	8	6.2	1.27	0.45	1.33	0.61	2.52	1612	0.87
17p12_CMT1A_dup	renal	374276	113	7292	3	2.2	1.35	0.45	1.55	0.43	3.88	1613	0.87
17p12_CMT1A_dup	hypertension	260904	83	120664	33	38.4	0.90	0.48	0.86	0.56	1.30	1690	0.88
17p12_CMT1A_dup	heart_valve	373191	115	8377	1	2.6	0.39	0.48	0.59	0.07	2.13	1704	0.88
17p12_CMT1A_dup	coagulation_defects	379026	115	2542	1	0.8	1.29	0.49	1.87	0.21	6.74	1735	0.89
17p12_CMT1A_dup	diabetes_other	360826	108	20742	8	6.2	1.27	0.51	1.28	0.58	2.46	1796	0.89
17p12_CMT1A_dup	diabetes_insulin_dependent	378973	115	2595	1	0.8	1.27	0.56	1.69	0.19	6.12	1910	0.91
17p12_CMT1A_dup	migraine	367885	111	13683	5	4.1	1.20	0.56	1.30	0.48	2.81	1927	0.91
17p12_CMT1A_dup	ovarian_cysts	374179	115	7389	1	2.3	0.45	0.57	0.65	0.07	2.37	1940	0.92
17p12_CMT1A_dup	nasal	378375	115	3193	1	1.0	1.03	0.59	1.60	0.18	5.80	1996	0.93
17p12_CMT1A_dup	high_cholesterol	314966	94	66602	22	19.9	1.09	0.60	1.14	0.69	1.81	2020	0.94
17p12_CMT1A_dup	died_to_2016	370595	112	10973	4	3.3	1.20	0.63	1.28	0.42	2.94	2079	0.94
17p12_CMT1A_dup	thyroid	354419	107	27149	9	8.2	1.09	0.63	1.19	0.56	2.22	2088	0.94
17p12_CMT1A_dup	biliary	360787	109	20781	7	6.3	1.11	0.64	1.20	0.52	2.36	2116	0.94
17p12_CMT1A_dup	low_WBC_count	378603	116	2965	0	0.9	0.00	0.65	0.56	0.00	3.80	2140	0.95
17p12_CMT1A_dup	atherosclerotic_vascular_disease	375817	114	5751	2	1.7	1.14	0.66	1.35	0.28	3.90	2165	0.95
17p12_CMT1A_dup	heart_other	369743	112	11825	4	3.6	1.11	0.67	1.24	0.41	2.86	2201	0.95
17p12_CMT1A_dup	hepatic	372812	113	8756	3	2.7	1.13	0.69	1.25	0.35	3.14	2265	0.95
17p12_CMT1A_dup	glaucoma	375039	115	6529	1	2.0	0.50	0.72	0.75	0.09	2.73	2326	0.96
17p12_CMT1A_dup	psoriasis	375547	115	6021	1	1.8	0.55	0.77	0.80	0.09	2.88	2505	0.97
17p12_CMT1A_dup	irritable_bowel_syndrome	368562	113	13006	3	4.0	0.76	0.80	0.87	0.24	2.19	2585	0.97
17p12_CMT1A_dup	Ischaemic heart disease not MI	365241	112	16327	4	5.0	0.81	0.81	0.89	0.29	2.07	2609	0.97
17p12_CMT1A_dup	diverticular_disease_intestine	354427	108	27141	8	8.3	0.97	0.85	1.07	0.49	2.05	2727	0.97
17p12_CMT1A_dup	obesity	371728	113	9840	3	3.0	1.00	0.92	1.06	0.29	2.65	2924	0.98
17p12_CMT1A_dup	uterine_problem	339268	104	42300	12	13.0	0.93	0.94	0.98	0.50	1.76	2976	0.99
17p12_CMT1A_dup	cataract	359712	110	21856	6	6.7	0.90	0.98	0.99	0.40	2.06	3094	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
17p12 HNPP_del	neuropathies	365743	194	15928	25	8.4	2.74	5.44E-06	3.02	1.95	4.49	10	0.0017
17p12 HNPP_del	diabetes_other	360915	197	20756	22	11.3	1.85	0.0061	1.97	1.23	3.00	87	0.22
17p12 HNPP_del	hernia	333767	203	47904	16	29.1	0.58	0.0089	0.54	0.31	0.86	111	0.25
17p12 HNPP_del	varicose veins	369817	217	11854	2	7.0	0.29	0.049	0.35	0.07	1.00	325	0.47
17p12 HNPP_del	any_Ca	331955	199	49716	20	29.8	0.70	0.053	0.65	0.40	1.00	340	0.49
17p12 HNPP_del	died_to_2016	370700	217	10971	2	6.4	0.32	0.071	0.38	0.08	1.07	412	0.54
17p12 HNPP_del	congenital	376644	219	5027	0	2.9	0.00	0.083	0.17	0.00	1.17	464	0.56
17p12 HNPP_del	ear	367439	206	14232	13	8.0	1.59	0.098	1.65	0.90	2.75	523	0.59
17p12 HNPP_del	renal_failure	373743	211	7928	8	4.5	1.76	0.12	1.83	0.84	3.43	580	0.63
17p12 HNPP_del	osteoarthritis	312933	186	68738	33	40.9	0.84	0.15	0.76	0.52	1.10	694	0.68
17p12 HNPP_del	hypertension	260960	139	120711	80	64.3	1.16	0.18	1.22	0.91	1.62	793	0.73
17p12 HNPP_del	anaemia	362516	204	19155	15	10.8	1.37	0.19	1.44	0.82	2.34	808	0.74
17p12 HNPP_del	ureter_bladder	333122	197	48549	22	28.7	0.79	0.20	0.76	0.48	1.15	860	0.75
17p12 HNPP_del	irritable_bowel_syndrome	368664	215	13007	4	7.6	0.54	0.24	0.60	0.20	1.35	961	0.78
17p12 HNPP_del	respiratory	360913	211	20758	8	12.1	0.67	0.25	0.68	0.32	1.27	993	0.78
17p12 HNPP_del	nasal	378476	216	3195	3	1.8	1.64	0.28	1.89	0.53	4.69	1105	0.80
17p12 HNPP_del	venous_thromboembolic_disease	367694	214	13977	5	8.1	0.62	0.30	0.66	0.25	1.40	1169	0.81
17p12 HNPP_del	diverticular_disease_intestine	354518	199	27153	20	15.2	1.28	0.32	1.27	0.78	1.97	1220	0.82
17p12 HNPP_del	thyroid	354512	200	27159	19	15.3	1.22	0.32	1.28	0.77	2.01	1238	0.82
17p12 HNPP_del	uterine_problem	339364	200	42307	19	24.9	0.78	0.32	0.79	0.47	1.25	1240	0.82
17p12 HNPP_del	gastric_reflux	347352	195	34319	24	19.3	1.22	0.35	1.23	0.79	1.84	1319	0.83
17p12 HNPP_del	gastrointestinal_ulcer	371311	211	10360	8	5.9	1.35	0.36	1.40	0.65	2.62	1354	0.84
17p12 HNPP_del	endocrine_other	379309	217	2362	2	1.4	1.48	0.38	1.85	0.39	5.25	1399	0.85
17p12 HNPP_del	osteoporosis	370069	210	11602	9	6.6	1.35	0.39	1.35	0.65	2.49	1460	0.85
17p12 HNPP_del	hepatic	372915	216	8756	3	5.1	0.60	0.45	0.68	0.19	1.69	1621	0.88
17p12 HNPP_del	diabetes_insulin_dependent	379075	217	2596	2	1.5	1.34	0.46	1.67	0.35	4.73	1634	0.88
17p12 HNPP_del	glaucoma	375138	214	6533	5	3.7	1.33	0.46	1.40	0.53	2.98	1636	0.88
17p12 HNPP_del	obesity	371827	212	9844	7	5.6	1.24	0.47	1.33	0.58	2.56	1660	0.88
17p12 HNPP_del	inflammatory_bowel_disease	363073	211	18598	8	10.8	0.75	0.47	0.78	0.36	1.46	1680	0.88
17p12 HNPP_del	gout	374576	214	7095	5	4.1	1.23	0.55	1.31	0.49	2.82	1883	0.91
17p12 HNPP_del	allergy	322981	189	58690	30	34.3	0.89	0.55	0.89	0.60	1.29	1889	0.91
17p12 HNPP_del	sciatica	338538	197	43133	22	25.1	0.89	0.56	0.88	0.55	1.33	1907	0.91
17p12 HNPP_del	atherosclerotic_vascular_disease	375920	217	5751	2	3.3	0.61	0.58	0.72	0.15	2.04	1965	0.93
17p12 HNPP_del	arrhythmia	357501	207	24170	12	14.0	0.87	0.59	0.86	0.46	1.46	1978	0.93
17p12 HNPP_del	cerebrovascular_disease_not_stroke	377338	216	4333	3	2.5	1.21	0.62	1.33	0.37	3.30	2056	0.94
17p12 HNPP_del	connective_tissue_disorder	372313	213	9358	6	5.4	1.12	0.69	1.18	0.48	2.38	2266	0.95
17p12 HNPP_del	stroke_derived2017	372849	215	8822	4	5.1	0.79	0.72	0.85	0.28	1.93	2370	0.96
17p12 HNPP_del	prostate_hyperplasia	367215	210	14456	9	8.3	1.09	0.74	1.12	0.53	2.12	2413	0.97
17p12 HNPP_del	paralytic_syndromes	379710	218	1961	1	1.1	0.89	0.75	1.32	0.15	4.73	2419	0.97
17p12 HNPP_del	COPD	367433	209	14238	10	8.1	1.22	0.77	1.11	0.53	2.03	2493	0.97
17p12 HNPP_del	aneurism	379704	218	1967	1	1.1	0.89	0.78	1.27	0.15	4.59	2520	0.97
17p12 HNPP_del	ovarian_cysts	374280	216	7391	3	4.3	0.71	0.79	0.87	0.24	2.17	2549	0.97
17p12 HNPP_del	high_cholesterol	315050	178	66621	41	37.6	1.07	0.80	1.05	0.73	1.48	2575	0.97
17p12 HNPP_del	heartfailure	376593	216	5078	3	2.9	1.03	0.80	1.15	0.32	2.86	2597	0.97
17p12 HNPP_del	heart_valve	373290	214	8381	5	4.8	1.04	0.81	1.11	0.42	2.35	2618	0.97
17p12 HNPP_del	digestive	299778	170	81893	49	46.4	1.04	0.81	1.04	0.75	1.42	2622	0.97
17p12 HNPP_del	heart_other	369844	213	11827	6	6.8	0.88	0.84	0.92	0.38	1.87	2718	0.97
17p12 HNPP_del	ocular	359998	206	21673	13	12.4	1.05	0.85	1.05	0.58	1.76	2738	0.97
17p12 HNPP_del	biliary	360886	208	20785	11	12.0	0.92	0.85	0.95	0.49	1.64	2745	0.97
17p12 HNPP_del	low_WBC_count	378705	218	2966	1	1.7	0.59	0.87	0.88	0.10	3.14	2786	0.98
17p12 HNPP_del	ischaemic_heart_disease_not_MI	365339	210	16332	9	9.4	0.96	0.89	0.96	0.46	1.74	2844	0.98
17p12 HNPP_del	renal	374378	215	7293	4	4.2	0.96	0.90	1.07	0.36	2.41	2866	0.98
17p12 HNPP_del	asthma	332320	192	49351	27	28.5	0.95	0.91	0.98	0.64	1.43	2904	0.98
17p12 HNPP_del	cataract	359808	206	21863	13	12.5	1.04	0.93	0.98	0.53	1.65	2961	0.99
17p12 HNPP_del	migraine	367986	212	13685	7	7.9	0.89	0.94	0.97	0.43	1.88	2996	0.99
17p12 HNPP_del	MI_derived2017	367803	211	13868	8	8.0	1.01	0.96	1.02	0.47	1.92	3040	0.99
17p12 HNPP_del	coagulation_defects	379129	218	2542	1	1.5	0.69	0.98	1.02	0.12	3.65	3090	1.00
17p12 HNPP_del	psoriasis	375648	216	6023	3	3.5	0.87	1.00	1.00	0.28	2.48	3121	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
17q11.2_del_NF1	diverticular_disease_intestine	354325	6	27136	3	0.5	4.69	0.022	5.66	1.32	20.54	203	0.34
17q11.2_del_NF1	congenital	376433	8	5028	1	0.1	8.43	0.027	14.03	1.49	62.26	225	0.37
17q11.2_del_NF1	ovarian_cysts	374072	8	7389	1	0.2	5.74	0.032	13.67	1.32	83.50	255	0.40
17q11.2_del_NF1	anaemia	362319	7	19142	2	0.4	4.43	0.035	6.28	1.17	23.69	266	0.41
17q11.2_del_NF1	digestive	299612	4	81849	5	1.1	2.59	0.037	3.87	1.09	14.46	273	0.42
17q11.2_del_NF1	thyroid	354319	7	27142	2	0.5	3.12	0.071	4.82	0.85	19.76	411	0.54
17q11.2_del_NF1	hypertension	260829	8	120632	1	0.7	0.35	0.094	0.26	0.03	1.22	501	0.59
17q11.2_del_NF1	high_cholesterol	314881	9	66580	0	1.9	0.00	0.098	0.16	0.00	1.30	521	0.59
17q11.2_del_NF1	uterine_problem	339171	7	42290	2	0.9	2.00	0.13	4.14	0.64	26.87	625	0.64
17q11.2_del_NF1	paralytic_syndromes	379501	9	1960	0	0.0	0.00	0.25	9.50	0.07	75.82	998	0.78
17q11.2_del_NF1	asthma	332135	7	49326	2	1.0	1.72	0.26	2.43	0.45	9.10	1038	0.79
17q11.2_del_NF1	endocrine_other	379101	9	2360	0	0.1	0.00	0.27	8.20	0.06	65.16	1064	0.80
17q11.2_del_NF1	biliary	360686	8	20775	1	0.5	2.04	0.28	3.01	0.32	13.64	1081	0.80
17q11.2_del_NF1	coagulation_defects	378920	9	2541	0	0.1	0.00	0.29	7.26	0.06	57.46	1138	0.81
17q11.2_del_NF1	aneurism	379495	9	1966	0	0.0	0.00	0.30	7.10	0.05	60.66	1163	0.81
17q11.2_del_NF1	diabetes_insulin_dependent	378867	9	2594	0	0.1	0.00	0.30	6.99	0.05	56.49	1167	0.81
17q11.2_del_NF1	low_WBC_count	378496	9	2965	0	0.1	0.00	0.32	6.35	0.05	50.20	1231	0.82
17q11.2_del_NF1	respiratory	360710	8	20751	1	0.5	2.04	0.33	2.67	0.28	11.99	1242	0.82
17q11.2_del_NF1	nasal	378269	9	3192	0	0.1	0.00	0.36	5.21	0.04	42.14	1351	0.85
17q11.2_del_NF1	ocular	359800	8	21661	1	0.5	1.96	0.36	2.45	0.26	11.01	1353	0.84
17q11.2_del_NF1	allergy	322799	7	58662	2	1.3	1.45	0.39	1.95	0.36	7.33	1444	0.85
17q11.2_del_NF1	sciatica	338350	9	43111	0	1.1	0.00	0.44	0.38	0.00	3.02	1588	0.87
17q11.2_del_NF1	prostate_hyperplasia	367013	8	14448	1	0.3	2.93	0.44	2.22	0.21	13.28	1596	0.87
17q11.2_del_NF1	hernia	333571	7	47890	2	1.0	1.77	0.44	1.82	0.33	7.11	1597	0.87
17q11.2_del_NF1	any_Ca	331763	7	49698	2	1.0	1.71	0.46	1.78	0.33	6.79	1640	0.88
17q11.2_del_NF1	arrhythmia	357302	8	24159	1	0.5	1.75	0.47	2.02	0.21	9.30	1671	0.88
17q11.2_del_NF1	cerebrovascular_disease_not_stroke	377131	9	4330	0	0.1	0.00	0.48	3.37	0.03	27.49	1708	0.88
17q11.2_del_NF1	psoriasis	375441	9	6020	0	0.1	0.00	0.50	3.17	0.02	25.07	1752	0.89
17q11.2_del_NF1	heartfailure	376386	9	5075	0	0.1	0.00	0.53	2.91	0.02	24.34	1838	0.90
17q11.2_del_NF1	atherosclerotic_vascular_disease	375712	9	5749	0	0.1	0.00	0.54	2.77	0.02	22.46	1873	0.91

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVS	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
17q12_del	diabetes_insulin_dependent	378861	3	2598	4	0.0	84.03	3.93E-08	135.87	31.15	641.08	4	3.08E-05
17q12_del	digestive	299609	1	81850	6	0.3	3.99	0.00044	15.44	3.17	150.30	33	0.042
17q12_del	renal	374168	5	7291	2	0.1	14.95	0.0021	20.82	3.71	87.49	57	0.11
17q12_del	low_WBC_count	378493	6	2966	1	0.0	18.38	0.0072	32.49	3.38	154.82	99	0.23
17q12_del	paralytic_syndromes	379498	6	1961	1	0.0	27.80	0.0083	30.14	3.09	148.20	103	0.25
17q12_del	coagulation_defects	378917	6	2542	1	0.0	21.45	0.0089	28.68	2.96	138.21	112	0.25
17q12_del	cataract	359607	5	21852	2	0.3	4.99	0.014	10.31	1.73	47.81	149	0.30
17q12_del	cerebrovascular_disease_not_stroke	377128	6	4331	1	0.1	12.59	0.017	20.09	2.00	105.62	168	0.31
17q12_del	ocular	359797	5	21662	2	0.3	5.03	0.023	7.90	1.39	33.67	207	0.35
17q12_del	glaucoma	374930	6	6529	1	0.1	8.35	0.027	14.70	1.48	75.86	230	0.37
17q12_del	ureter_bladder	332929	4	48530	3	0.6	3.37	0.038	5.08	1.11	21.55	277	0.43
17q12_del	hernia	333568	4	47891	3	0.6	3.41	0.054	4.62	0.97	20.71	347	0.48
17q12_del	renal_failure	373538	6	7921	1	0.1	6.88	0.061	8.69	0.88	44.55	368	0.52
17q12_del	varicose_veins	369606	6	11853	1	0.2	4.60	0.066	8.04	0.83	38.55	381	0.55
17q12_del	hepatic	372705	6	8754	1	0.1	6.23	0.075	7.45	0.77	36.18	429	0.55
17q12_del	gout	374368	6	7091	1	0.1	7.69	0.079	7.42	0.74	39.84	449	0.55
17q12_del	died_to_2016	370489	6	10970	1	0.2	4.97	0.11	5.90	0.61	28.99	544	0.62
17q12_del	biliary	360684	6	20775	1	0.3	2.62	0.13	5.05	0.52	24.31	652	0.65
17q12_del	high_cholesterol	314876	4	66583	3	0.8	2.46	0.15	3.18	0.64	15.41	690	0.68
17q12_del	ischaemic_heart_disease_not_MI	365135	6	16324	1	0.3	3.34	0.18	4.30	0.43	22.72	773	0.72
17q12_del	inflammatory_bowel_disease	362868	6	18591	1	0.3	2.93	0.18	4.12	0.43	19.70	780	0.73
17q12_del	asthma	332133	5	49326	2	0.7	2.21	0.18	3.03	0.54	12.60	785	0.73
17q12_del	MI_derived2017	367598	6	13861	1	0.2	3.93	0.20	4.02	0.40	21.75	834	0.75
17q12_del	endocrine_other	379099	7	2360	0	0.0	0.00	0.22	11.52	0.09	95.08	903	0.76
17q12_del	aneurism	379493	7	1966	0	0.0	0.00	0.24	10.43	0.08	94.86	952	0.78
17q12_del	respiratory	360708	6	20751	1	0.3	2.63	0.26	3.19	0.33	15.74	1037	0.79
17q12_del	nasal	378267	7	3192	0	0.1	0.00	0.28	7.90	0.06	66.90	1096	0.80
17q12_del	diabetes_other	360724	6	20735	1	0.3	2.63	0.32	2.78	0.28	14.39	1234	0.82
17q12_del	osteoarthritis	312754	7	68705	0	1.5	0.00	0.38	0.33	0.00	2.85	1408	0.85
17q12_del	congenital	376432	7	5027	0	0.1	0.00	0.39	4.66	0.04	38.37	1452	0.85
17q12_del	osteoporosis	369866	7	11593	0	0.2	0.00	0.40	4.55	0.03	38.40	1470	0.85
17q12_del	ovarian_cysts	374071	7	7388	0	0.1	0.00	0.43	4.23	0.03	52.32	1546	0.87
17q12_del	gastric_reflux	347163	6	34296	1	0.6	1.59	0.43	2.18	0.22	10.53	1556	0.87
17q12_del	psoriasis	375439	7	6020	0	0.1	0.00	0.47	3.55	0.03	29.39	1657	0.88
17q12_del	allergy	322799	7	58660	0	1.3	0.00	0.47	0.40	0.00	3.29	1673	0.88
17q12_del	heartfailure	376384	7	5075	0	0.1	0.00	0.48	3.44	0.03	31.02	1703	0.88
17q12_del	connective_tissue_disorder	372107	7	9352	0	0.2	0.00	0.49	3.25	0.02	26.96	1739	0.89
17q12_del	atherosclerotic_vascular_disease	375710	7	5749	0	0.1	0.00	0.51	3.14	0.02	27.21	1775	0.89
17q12_del	heart_valve	373083	7	8376	0	0.2	0.00	0.55	2.75	0.02	23.35	1885	0.91
17q12_del	any_Ca	331762	6	49697	1	0.9	1.10	0.56	1.76	0.18	8.68	1925	0.91
17q12_del	sciatica	338347	6	43112	1	0.8	1.26	0.61	1.62	0.17	7.78	2047	0.94
17q12_del	obesity	371622	7	9837	0	0.2	0.00	0.62	2.24	0.02	18.62	2070	0.94
17q12_del	stroke_derived2017	372641	7	8818	0	0.2	0.00	0.63	2.19	0.02	19.06	2107	0.94
17q12_del	migraine	367781	7	13678	0	0.3	0.00	0.65	2.09	0.02	18.51	2157	0.95
17q12_del	irritable_bowel_syndrome	368456	7	13003	0	0.2	0.00	0.65	2.08	0.02	17.69	2159	0.95
17q12_del	gastrointestinal_ulcer	371107	7	10352	0	0.2	0.00	0.72	1.79	0.01	15.28	2324	0.96
17q12_del	venous_thromboembolic_disease	367487	7	13972	0	0.3	0.00	0.74	1.71	0.01	14.26	2394	0.96
17q12_del	neuropathies	365556	7	15903	0	0.3	0.00	0.76	1.63	0.01	13.43	2455	0.96
17q12_del	heart_other	369638	7	11821	0	0.2	0.00	0.76	1.61	0.01	14.05	2472	0.97
17q12_del	ear	367240	7	14219	0	0.3	0.00	0.77	1.58	0.01	13.21	2491	0.97
17q12_del	hypertension	260826	5	120633	2	2.3	0.90	0.80	0.82	0.14	3.72	2586	0.97
17q12_del	uterine_problem	339171	7	42288	0	0.9	0.00	0.82	0.71	0.01	8.71	2633	0.97
17q12_del	thyroid	354319	7	27140	0	0.5	0.00	0.83	1.41	0.01	12.20	2658	0.97
17q12_del	COPD	367231	7	14228	0	0.3	0.00	0.83	1.38	0.01	12.35	2683	0.97
17q12_del	anaemia	362319	7	19140	0	0.4	0.00	0.88	1.25	0.01	10.31	2817	0.98
17q12_del	arrhythmia	357301	7	24158	0	0.5	0.00	0.92	0.86	0.01	7.50	2926	0.98
17q12_del	prostate_hyperplasia	367012	7	14447	0	0.3	0.00	0.96	0.93	0.01	8.50	3034	0.99
17q12_del	diverticular_disease_intestine	354326	7	27133	0	0.5	0.00	0.96	0.93	0.01	8.04	3047	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
17q12_dup	renal_failure	373622	90	7929	9	1.9	4.38	0.00029	4.56	2.16	8.57	26	0.035
17q12_dup	prostate_hyperplasia	367092	87	14459	12	3.4	3.20	0.0044	2.84	1.41	5.33	75	0.19
17q12_dup	respiratory	360790	88	20761	11	5.1	2.04	0.034	2.08	1.06	3.72	260	0.41
17q12_dup	renal	374257	94	7294	5	1.8	2.64	0.035	2.93	1.09	6.31	268	0.41
17q12_dup	died_to_2016	370575	92	10976	7	2.7	2.46	0.040	2.42	1.04	4.78	295	0.43
17q12_dup	connective_tissue_disorder	372194	94	9357	5	2.4	2.06	0.056	2.62	0.97	5.67	355	0.50
17q12_dup	paralytic_syndromes	379589	97	1962	2	0.5	3.93	0.063	4.35	0.90	12.53	372	0.53
17q12_dup	glaucoma	375019	95	6532	4	1.7	2.36	0.076	2.70	0.89	6.29	431	0.55
17q12_dup	coagulation_defects	379008	97	2543	2	0.7	3.03	0.096	3.62	0.75	10.39	508	0.59
17q12_dup	asthma	332209	81	49342	18	12.0	1.41	0.11	1.54	0.90	2.49	558	0.62
17q12_dup	digestive	299679	71	81872	28	19.4	1.32	0.12	1.44	0.91	2.21	578	0.63
17q12_dup	hepatic	372798	99	8753	0	2.3	0.00	0.12	0.19	0.00	1.34	584	0.63
17q12_dup	biliary	360769	91	20782	8	5.2	1.48	0.12	1.84	0.84	3.52	596	0.63
17q12_dup	diverticular_disease_intestine	354415	96	27136	3	7.4	0.43	0.13	0.48	0.13	1.21	642	0.64
17q12_dup	hypertension	260881	60	120670	39	27.8	1.25	0.15	1.37	0.89	2.09	684	0.68
17q12_dup	ovarian_cysts	374160	96	7391	3	1.9	1.56	0.16	2.45	0.66	6.44	713	0.69
17q12_dup	arrhythmia	357383	89	24168	10	6.0	1.59	0.16	1.63	0.80	2.99	731	0.70
17q12_dup	heart_valve	373171	95	8380	4	2.1	1.84	0.18	2.05	0.67	4.73	783	0.73
17q12_dup	ear	367326	93	14225	6	3.6	1.63	0.19	1.77	0.72	3.63	822	0.74
17q12_dup	diabetes_other	360808	90	20743	9	5.2	1.67	0.20	1.61	0.76	3.02	840	0.74
17q12_dup	ureter_bladder	333007	82	48544	17	12.0	1.35	0.20	1.42	0.82	2.34	852	0.74
17q12_dup	irritable_bowel_syndrome	368543	94	13008	5	3.3	1.48	0.21	1.84	0.68	3.98	862	0.75
17q12_dup	high_cholesterol	314957	85	66594	14	18.0	0.81	0.21	0.70	0.38	1.21	869	0.75
17q12_dup	psoriasis	375528	96	6023	3	1.5	1.92	0.22	2.10	0.58	5.28	906	0.77
17q12_dup	COPD	367317	93	14234	6	3.6	1.62	0.23	1.71	0.69	3.56	916	0.77
17q12_dup	thyroid	354403	91	27148	8	7.0	1.14	0.24	1.58	0.72	3.07	953	0.78
17q12_dup	anaemia	362404	92	19147	7	4.9	1.41	0.25	1.60	0.69	3.16	987	0.78
17q12_dup	atherosclerotic_vascular_disease	375802	99	5749	0	1.5	0.00	0.28	0.29	0.00	2.00	1080	0.80
17q12_dup	migraine	367872	98	13679	1	3.6	0.28	0.28	0.46	0.05	1.66	1082	0.80
17q12_dup	heart_other	369725	94	11826	5	3.0	1.63	0.31	1.62	0.60	3.52	1191	0.81
17q12_dup	endocrine_other	379190	98	2361	1	0.6	1.63	0.31	2.62	0.30	9.50	1198	0.81
17q12_dup	gastric_reflux	374250	93	34301	6	9.2	0.67	0.35	0.70	0.28	1.43	1316	0.83
17q12_dup	sciatica	338426	85	43125	14	10.8	1.25	0.35	1.32	0.72	2.23	1317	0.83
17q12_dup	congenital	376522	97	5029	2	1.3	1.53	0.37	1.89	0.39	5.43	1356	0.84
17q12_dup	ocular	359884	92	21667	7	5.5	1.25	0.43	1.37	0.59	2.71	1552	0.87
17q12_dup	low_WBC_count	378585	98	2966	1	0.8	1.30	0.43	2.06	0.24	7.48	1553	0.87
17q12_dup	osteoporosis	369955	96	11596	3	3.0	1.00	0.44	1.58	0.43	4.07	1581	0.87
17q12_dup	diabetes_insulin_dependent	378956	98	2595	1	0.7	1.49	0.48	1.91	0.22	6.94	1699	0.88
17q12_dup	uterine_problem	339257	93	42294	6	11.6	0.55	0.48	0.75	0.29	1.62	1707	0.88
17q12_dup	osteoarthritis	312827	80	68724	19	17.6	1.07	0.50	1.20	0.70	1.95	1753	0.89
17q12_dup	allergy	322879	87	58672	12	15.8	0.79	0.51	0.82	0.43	1.43	1783	0.89
17q12_dup	gout	374458	96	7093	3	1.8	1.63	0.52	1.45	0.40	3.70	1826	0.90
17q12_dup	nasal	378358	98	3193	1	0.8	1.21	0.54	1.73	0.20	6.28	1861	0.91
17q12_dup	stroke_derived2017	372730	96	8821	3	2.3	1.31	0.56	1.40	0.39	3.54	1914	0.91
17q12_dup	obesity	371711	96	9840	3	2.5	1.18	0.61	1.34	0.37	3.36	2043	0.94
17q12_dup	hernia	333648	84	47903	15	12.1	1.21	0.61	1.16	0.64	1.95	2052	0.94
17q12_dup	cataract	359695	93	21856	6	5.7	1.06	0.67	1.20	0.48	2.52	2197	0.95
17q12_dup	inflammatory_bowel_disease	362956	94	18595	5	4.8	1.04	0.77	1.14	0.42	2.45	2504	0.97
17q12_dup	cerebrovascular_disease_not_stroke	377220	98	4331	1	1.1	0.89	0.78	1.27	0.14	4.62	2535	0.97
17q12_dup	gastrointestinal_ulcer	371196	96	10355	3	2.7	1.12	0.78	1.17	0.32	2.94	2542	0.97
17q12_dup	varicose_veins	369697	97	11854	2	3.1	0.65	0.80	0.85	0.18	2.44	2582	0.97
17q12_dup	neuropathies	365645	96	15906	3	4.2	0.73	0.82	0.89	0.25	2.23	2653	0.97
17q12_dup	aneurism	379585	99	1966	0	0.5	0.00	0.89	0.82	0.01	5.69	2830	0.98
17q12_dup	ischaemic_heart_disease_not_MI	365224	95	16327	4	4.2	0.94	0.91	0.94	0.31	2.20	2890	0.98
17q12_dup	heartfailure	376475	98	5076	1	1.3	0.76	0.95	0.95	0.11	3.47	3001	0.99
17q12_dup	any_Ca	331843	87	49708	12	13.0	0.93	0.95	0.98	0.51	1.72	3002	0.99
17q12_dup	venous_thromboembolic_disease	367576	96	13975	3	3.6	0.83	0.95	0.96	0.27	2.43	3004	0.99
17q12_dup	MI_derived2017	367687	95	13864	4	3.6	1.11	1.00	1.00	0.33	2.34	3126	1.00



CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
1q21.1_del	cataract	359694	92	21864	14	5.6	2.31	0.00047	3.23	1.74	5.60	35	0.042
1q21.1_del	heartfailure	376477	100	5081	6	1.3	4.25	0.0011	5.34	2.14	11.15	40	0.083
1q21.1_del	COPD	367319	95	14239	11	3.7	2.78	0.0021	3.13	1.57	5.69	56	0.11
1q21.1_del	congenital	376526	101	5032	5	1.3	3.58	0.0098	3.92	1.46	8.42	122	0.25
1q21.1_del	arrhythmia	357388	94	24170	12	6.4	1.79	0.022	2.16	1.13	3.80	198	0.35
1q21.1_del	osteoarthritis	312826	79	68732	27	17.4	1.41	0.026	1.70	1.07	2.64	223	0.36
1q21.1_del	ocular	359887	95	21671	11	5.7	1.83	0.028	2.14	1.09	3.80	235	0.37
1q21.1_del	died_to_2016	370582	99	10976	7	2.9	2.30	0.031	2.53	1.10	5.00	250	0.39
1q21.1_del	heart_other	369731	100	11827	6	3.2	1.83	0.084	2.20	0.89	4.55	467	0.56
1q21.1_del	endocrine_other	379196	104	2362	2	0.6	3.05	0.084	3.84	0.80	11.02	468	0.56
1q21.1_del	inflammatory_bowel_disease	362959	97	18599	9	5.0	1.74	0.089	1.88	0.90	3.48	487	0.57
1q21.1_del	gastric_reflux	347249	92	34309	14	9.1	1.47	0.11	1.61	0.88	2.72	573	0.63
1q21.1_del	connective_tissue_disorder	372201	101	9357	5	2.5	1.92	0.11	2.17	0.80	4.70	574	0.63
1q21.1_del	ischaemic_heart_disease_not_MI	365228	99	16330	7	4.4	1.54	0.14	1.86	0.80	3.72	663	0.65
1q21.1_del	asthma	332215	87	49343	19	12.9	1.39	0.15	1.46	0.87	2.33	685	0.68
1q21.1_del	high_cholesterol	314966	94	66592	12	19.9	0.65	0.15	0.65	0.34	1.16	692	0.68
1q21.1_del	ovarian_cysts	374170	106	7388	0	2.1	0.00	0.15	0.22	0.00	1.50	701	0.69
1q21.1_del	gastrointestinal_ulcer	371201	101	10357	5	2.8	1.74	0.16	1.99	0.74	4.32	712	0.69
1q21.1_del	heart_valve	373178	102	8380	4	2.3	1.72	0.17	2.09	0.69	4.81	750	0.71
1q21.1_del	hernia	333653	89	47905	17	12.8	1.28	0.17	1.46	0.84	2.40	759	0.72
1q21.1_del	neuropathies	365648	99	15910	7	4.3	1.58	0.20	1.70	0.74	3.34	823	0.74
1q21.1_del	ureter_bladder	333014	89	48544	17	13.0	1.26	0.21	1.41	0.82	2.31	870	0.75
1q21.1_del	osteoporosis	369960	101	11598	5	3.2	1.55	0.21	1.85	0.67	4.11	874	0.75
1q21.1_del	sciatica	338431	90	43127	16	11.5	1.34	0.23	1.40	0.80	2.31	918	0.77
1q21.1_del	aneurism	379591	105	1967	1	0.5	1.83	0.26	3.01	0.34	11.09	1021	0.79
1q21.1_del	digestive	299687	79	81871	27	21.6	1.19	0.26	1.29	0.82	1.98	1032	0.79
1q21.1_del	prostate_hyperplasia	367106	101	14452	5	4.0	1.25	0.27	1.74	0.61	4.06	1062	0.80
1q21.1_del	respiratory	360805	103	20753	3	5.9	0.52	0.29	0.59	0.16	1.48	1130	0.81
1q21.1_del	renal	374266	103	7292	3	2.0	1.48	0.34	1.75	0.49	4.39	1304	0.83
1q21.1_del	uterine_problem	339255	91	42303	15	11.3	1.28	0.35	1.32	0.72	2.31	1326	0.84
1q21.1_del	renal_failure	373635	103	7923	3	2.2	1.36	0.37	1.70	0.47	4.31	1370	0.85
1q21.1_del	migraine	367878	104	13680	2	3.9	0.53	0.42	0.62	0.13	1.78	1519	0.87
1q21.1_del	coagulation_defects	379016	105	2542	1	0.7	1.42	0.44	2.05	0.23	7.41	1561	0.87
1q21.1_del	diabetes_insulin_dependent	378963	105	2595	1	0.7	1.39	0.45	1.99	0.23	7.22	1617	0.88
1q21.1_del	any_Ca	331852	96	49706	10	14.4	0.72	0.48	0.80	0.39	1.45	1715	0.88
1q21.1_del	cerebrovascular_disease_not_stroke	377228	106	4330	0	1.2	0.00	0.51	0.45	0.00	3.08	1794	0.90
1q21.1_del	stroke_derived2017	372739	105	8819	1	2.5	0.41	0.54	0.62	0.07	2.27	1850	0.91
1q21.1_del	allergy	322884	92	58674	14	16.7	0.86	0.55	0.85	0.47	1.43	1886	0.91
1q21.1_del	hepatic	372802	103	8756	3	2.4	1.23	0.56	1.40	0.39	3.52	1905	0.91
1q21.1_del	diabetes_other	360820	102	20738	4	5.9	0.69	0.59	0.78	0.26	1.80	1997	0.93
1q21.1_del	varicose_veins	369704	104	11854	2	3.3	0.61	0.61	0.73	0.15	2.09	2028	0.94
1q21.1_del	gout	374466	104	7092	2	2.0	1.02	0.62	1.40	0.29	4.10	2068	0.94
1q21.1_del	MI_derived2017	367694	102	13864	4	3.8	1.04	0.63	1.28	0.42	3.00	2087	0.94
1q21.1_del	nasal	378366	106	3192	0	0.9	0.00	0.67	0.58	0.00	3.95	2204	0.95
1q21.1_del	thyroid	354412	100	27146	6	7.7	0.80	0.68	0.85	0.34	1.76	2235	0.95
1q21.1_del	anaemia	362412	100	19146	6	5.3	1.13	0.69	1.18	0.48	2.42	2268	0.95
1q21.1_del	low_WBC_count	378593	106	2965	0	0.8	0.00	0.71	0.61	0.00	4.19	2307	0.96
1q21.1_del	irritable_bowel_syndrome	368551	102	13007	4	3.6	1.11	0.72	1.19	0.39	2.74	2361	0.96
1q21.1_del	obesity	371718	103	9840	3	2.7	1.10	0.77	1.18	0.33	2.97	2488	0.97
1q21.1_del	ear	367336	103	14222	3	4.0	0.76	0.80	0.87	0.24	2.19	2591	0.97
1q21.1_del	venous_thromboembolic_disease	367583	103	13975	3	3.9	0.77	0.85	0.91	0.25	2.27	2746	0.97
1q21.1_del	psoriasis	375537	105	6021	1	1.7	0.60	0.86	0.87	0.10	3.15	2772	0.98
1q21.1_del	diverticular_disease_intestine	354418	99	27140	7	7.6	0.93	0.87	1.07	0.46	2.12	2782	0.98
1q21.1_del	biliary	360779	101	20779	5	5.8	0.87	0.89	0.94	0.35	2.03	2834	0.98
1q21.1_del	glaucoma	375029	105	6529	1	1.8	0.55	0.91	0.91	0.10	3.31	2894	0.98
1q21.1_del	paralytic_syndromes	379598	106	1960	0	0.5	0.00	0.92	0.87	0.01	5.95	2918	0.98
1q21.1_del	atherosclerotic_vascular_disease	375808	105	5750	1	1.6	0.63	0.93	0.93	0.11	3.37	2942	0.98
1q21.1_del	hypertension	260895	74	120663	32	34.2	0.95	0.96	1.01	0.65	1.54	3029	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
1q21.1_dup	diabetes_other	360864	146	20756	22	8.4	2.41	0.00017	2.65	1.64	4.10	21	0.025
1q21.1_dup	heart_other	369787	156	11833	12	5.0	2.30	0.0056	2.55	1.35	4.39	82	0.21
1q21.1_dup	obesity	371772	157	9848	11	4.2	2.54	0.0086	2.50	1.29	4.36	108	0.25
1q21.1_dup	high_cholesterol	314999	127	66621	41	26.9	1.40	0.013	1.63	1.11	2.34	142	0.29
1q21.1_dup	irritable_bowel_syndrome	368605	156	13015	12	5.5	2.10	0.017	2.20	1.17	3.77	167	0.31
1q21.1_dup	hypertension	260923	102	120697	66	47.2	1.24	0.023	1.47	1.06	2.02	205	0.35
1q21.1_dup	hepatic	372858	159	8762	9	3.7	2.33	0.023	2.37	1.14	4.32	206	0.35
1q21.1_dup	diabetes_insulin_dependent	379022	164	2598	4	1.1	3.50	0.025	3.63	1.20	8.26	220	0.36
1q21.1_dup	respiratory	360854	152	20766	16	8.7	1.75	0.037	1.80	1.04	2.92	274	0.43
1q21.1_dup	died_to_2016	370641	158	10979	10	4.7	2.07	0.038	2.09	1.05	3.73	278	0.43
1q21.1_dup	anaemia	362465	153	19155	15	8.1	1.78	0.040	1.82	1.03	2.97	288	0.43
1q21.1_dup	stroke_derived2017	372794	160	8826	8	3.8	2.06	0.052	2.16	0.99	4.09	335	0.48
1q21.1_dup	biliary	360831	153	20789	15	8.8	1.64	0.055	1.74	0.99	2.86	351	0.49
1q21.1_dup	MI_derived2017	363750	158	13870	10	6.0	1.64	0.11	1.77	0.87	3.21	542	0.62
1q21.1_dup	inflammatory_bowel_disease	363017	155	18603	13	7.9	1.59	0.11	1.62	0.89	2.73	559	0.63
1q21.1_dup	digestive	299731	123	81889	45	33.6	1.25	0.12	1.32	0.93	1.85	589	0.63
1q21.1_dup	ischaemic_heart_disease_not_MI	365286	157	16334	11	7.0	1.53	0.13	1.67	0.85	2.94	621	0.64
1q21.1_dup	arrhythmia	357447	153	24173	15	10.3	1.41	0.13	1.54	0.87	2.54	633	0.64
1q21.1_dup	neuropathies	365706	157	15914	11	6.8	1.57	0.14	1.62	0.84	2.83	670	0.66
1q21.1_dup	allergy	322928	136	58692	32	24.7	1.24	0.17	1.32	0.88	1.91	756	0.72
1q21.1_dup	paralytic_syndromes	379658	166	1962	2	0.9	2.32	0.20	2.58	0.54	7.35	830	0.75
1q21.1_dup	gastric_reflux	347305	148	34315	20	14.6	1.32	0.20	1.37	0.84	2.13	838	0.74
1q21.1_dup	migraine	367933	159	13687	9	5.9	1.49	0.20	1.58	0.76	2.90	843	0.74
1q21.1_dup	COPD	367382	158	14238	10	6.1	1.60	0.20	1.55	0.77	2.80	861	0.75
1q21.1_dup	osteoporosis	370025	166	11595	2	5.2	0.39	0.22	0.50	0.10	1.42	898	0.76
1q21.1_dup	ureter_bladder	333067	142	48553	26	20.7	1.22	0.25	1.29	0.83	1.92	1010	0.78
1q21.1_dup	ovarian_cysts	374227	163	7393	5	3.2	1.54	0.29	1.65	0.61	3.56	1131	0.81
1q21.1_dup	prostate_hyperplasia	367165	160	14455	8	6.3	1.26	0.30	1.51	0.67	3.00	1156	0.81
1q21.1_dup	osteoarthritis	312890	143	68730	25	31.4	0.83	0.30	0.80	0.51	1.21	1172	0.81
1q21.1_dup	low_WBC_count	378653	166	2967	2	1.3	1.53	0.35	1.93	0.40	5.49	1314	0.83</

CNV	Phenotype	N_con	N_CNV_ con	N_case	N_CNV_ case	Expected_ N_CNVS	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
22q11.2_del	low_WBC_count	378496	9	2966	1	0.1	12.87	0.011	23.63	2.53	103.05	132	0.27
22q11.2_del	hypertension	260825	4	120637	6	1.9	1.90	0.020	4.61	1.28	18.00	176	0.35
22q11.2_del	COPD	367232	8	14230	2	0.3	5.36	0.025	7.60	1.35	30.44	218	0.36
22q11.2_del	heartfailure	376386	9	5076	1	0.1	7.52	0.038	11.90	1.19	59.49	281	0.43
22q11.2_del	heart_valve	373085	9	8377	1	0.2	4.55	0.053	9.22	0.97	41.69	338	0.49
22q11.2_del	diabetes_other	360726	8	20736	2	0.5	3.68	0.063	5.03	0.90	19.98	374	0.53
22q11.2_del	renal_failure	373541	9	7921	1	0.2	4.82	0.069	7.98	0.81	38.28	396	0.54
22q11.2_del	gastrointestinal_ulcer	371109	9	10353	1	0.3	3.68	0.11	5.76	0.60	26.30	549	0.62
22q11.2_del	prostate_hyperplasia	367014	9	14448	1	0.4	2.64	0.15	5.10	0.48	30.26	689	0.68
22q11.2_del	obesity	371624	9	9838	1	0.2	3.88	0.16	4.49	0.48	19.93	708	0.69
22q11.2_del	ear	367242	9	14220	1	0.3	2.68	0.16	4.45	0.47	19.62	714	0.69
22q11.2_del	asthma	332135	7	49327	3	1.0	2.32	0.16	2.67	0.65	9.03	721	0.69
22q11.2_del	ischaemic_heart_disease_not_MI	365138	9	16324	1	0.4	2.34	0.17	4.50	0.45	22.09	737	0.70
22q11.2_del	died_to_2016	370492	9	10970	1	0.3	3.48	0.17	4.21	0.44	18.98	752	0.72
22q11.2_del	ocular	359801	9	21661	1	0.5	1.76	0.22	3.50	0.37	15.61	911	0.77
22q11.2_del	biliary	360687	9	20775	1	0.5	1.84	0.23	3.47	0.37	15.31	917	0.77
22q11.2_del	diverticular_disease_intestine	354328	9	27134	1	0.7	1.41	0.25	3.29	0.34	15.12	991	0.78
22q11.2_del	aneurism	379496	10	1966	0	0.1	0.00	0.25	9.66	0.07	84.26	999	0.78
22q11.2_del	thyroid	354321	9	27141	1	0.7	1.41	0.26	3.23	0.34	14.97	1016	0.79
22q11.2_del	endocrine_other	379102	10	2360	0	0.1	0.00	0.27	8.10	0.06	63.52	1071	0.80
22q11.2_del	anemia	362321	9	19141	1	0.5	1.99	0.28	2.98	0.32	13.10	1087	0.80
22q11.2_del	ureter_bladder	332933	8	48529	2	1.2	1.57	0.28	2.35	0.44	8.78	1097	0.80
22q11.2_del	arrhythmia	357303	9	24159	1	0.6	1.58	0.29	2.92	0.30	13.60	1137	0.81
22q11.2_del	hernia	333572	8	47890	2	1.1	1.59	0.29	2.32	0.42	8.91	1141	0.81
22q11.2_del	nasal	378270	10	3192	0	0.1	0.00	0.31	6.69	0.05	52.94	1192	0.81
22q11.2_del	paralytic_syndromes	379502	10	1960	0	0.1	0.00	0.31	6.70	0.05	53.99	1193	0.81
22q11.2_del	coagulation_defects	378921	10	2541	0	0.1	0.00	0.32	6.51	0.05	51.00	1213	0.81
22q11.2_del	cerebrovascular_disease_not_stroke	377132	10	4330	0	0.1	0.00	0.34	5.95	0.05	50.34	1280	0.82
22q11.2_del	diabetes_insulin_dependent	378868	10	2594	0	0.1	0.00	0.38	4.88	0.04	39.19	1406	0.85
22q11.2_del	glaucoma	374934	10	6528	0	0.2	0.00	0.42	4.14	0.03	35.22	1536	0.87
22q11.2_del	osteoporosis	369869	10	11593	0	0.3	0.00	0.45	3.82	0.03	32.17	1602	0.87
22q11.2_del	uterine_problem	339174	10	42288	0	1.2	0.00	0.45	0.37	0.00	3.49	1616	0.87
22q11.2_del	high_cholesterol	314880	8	66582	2	1.7	1.15	0.46	1.84	0.32	7.57	1627	0.88
22q11.2_del	allergy	322800	8	58662	2	1.5	1.30	0.50	1.67	0.31	6.09	1765	0.89
22q11.2_del	congenital	376435	10	5027	0	0.1	0.00	0.52	2.96	0.02	23.06	1822	0.90
22q11.2_del	gastric_reflux	347166	9	34296	1	0.9	1.11	0.53	1.83	0.19	8.05	1830	0.90
22q11.2_del	atherosclerotic_vascular_disease	375713	10	5749	0	0.2	0.00	0.53	2.89	0.02	23.99	1847	0.90
22q11.2_del	psoriasis	375442	10	6020	0	0.2	0.00	0.58	2.48	0.02	19.42	1960	0.93
22q11.2_del	any_Ca	331766	10	49696	0	1.5	0.00	0.58	0.49	0.00	3.91	1969	0.93
22q11.2_del	connective_tissue_disorder	372110	10	9352	0	0.3	0.00	0.59	2.45	0.02	19.50	1982	0.93
22q11.2_del	renal	374173	10	7289	0	0.2	0.00	0.60	2.39	0.02	18.82	2000	0.93
22q11.2_del	ovarian_cysts	374074	10	7388	0	0.2	0.00	0.63	2.20	0.02	20.76	2110	0.94
22q11.2_del	gout	374372	10	7090	0	0.2	0.00	0.64	2.16	0.02	18.41	2118	0.94
22q11.2_del	stroke_derived2017	372644	10	8818	0	0.2	0.00	0.68	1.93	0.01	16.08	2250	0.95
22q11.2_del	varicose_veins	369610	10	11852	0	0.3	0.00	0.74	1.69	0.01	13.24	2399	0.96
22q11.2_del	hepatic	372709	10	8753	0	0.2	0.00	0.75	1.64	0.01	12.96	2440	0.97
22q11.2_del	heart_other	369641	10	11821	0	0.3	0.00	0.79	1.53	0.01	12.75	2546	0.97
22q11.2_del	cataract	359612	10	21850	0	0.6	0.00	0.79	1.53	0.01	13.51	2554	0.97
22q11.2_del	sciatica	338350	9	43112	1	1.1	0.88	0.81	1.24	0.13	5.42	2616	0.97
22q11.2_del	respiratory	360712	10	20750	0	0.6	0.00	0.82	0.73	0.01	5.84	2641	0.97
22q11.2_del	venous_thromboembolic_disease	367490	10	13972	0	0.4	0.00	0.83	1.38	0.01	10.91	2680	0.97
22q11.2_del	digestive	299616	8	81846	2	2.2	0.93	0.84	1.16	0.22	4.31	2693	0.97
22q11.2_del	irritable_bowel_syndrome	368459	10	13003	0	0.4	0.00	0.84	1.36	0.01	10.93	2701	0.97
22q11.2_del	migraine	367784	10	13678	0	0.4	0.00	0.90	1.22	0.01	9.98	2865	0.98
22q11.2_del	inflammatory_bowel_disease	362872	10	18590	0	0.5	0.00	0.93	0.88	0.01	6.92	2956	0.99
22q11.2_del	neuropsychopathies	365559	10	15903	0	0.4	0.00	0.94	1.12	0.01	8.81	2968	0.99
22q11.2_del	MI_derived2017	367602	10	13860	0	0.4	0.00	0.94	1.12	0.01	9.92	2978	0.99
22q11.2_del	osteoarthritis	312756	9	68706	1	2.0	0.56	0.97	1.03	0.11	4.73	3066	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
22q11.2_dup	gastric_reflux	347374	217	34344	49	21.5	2.05	1.82E-06	2.31	1.67	3.13	8	0.00071
22q11.2_dup	hernia	333779	215	47939	51	30.9	1.53	0.0013	1.71	1.24	2.32	45	0.094
22q11.2_dup	gout	374628	266	7090	0	5.0	0.00	0.0071	0.09	0.00	0.63	97	0.23
22q11.2_dup	ischaemic_heart_disease_not_MI	365375	246	16343	20	11.0	1.76	0.0087	1.96	1.20	3.03	109	0.25
22q11.2_dup	low_WBC_count	378747	260	2971	6	2.0	2.90	0.011	3.33	1.37	6.68	129	0.27
22q11.2_dup	heartfailure	376634	257	5084	9	3.5	2.54	0.011	2.64	1.27	4.83	133	0.27
22q11.2_dup	digestive	299799	191	81919	75	52.2	1.31	0.012	1.43	1.09	1.87	135	0.27
22q11.2_dup	MI_derived2017	367840	248	13878	18	9.4	1.86	0.014	1.95	1.16	3.10	147	0.29
22q11.2_dup	endocrine_other	379353	261	2365	5	1.6	3.04	0.017	3.41	1.28	7.21	171	0.32
22q11.2_dup	glaucoma	375181	257	6537	9	4.5	1.98	0.020	2.40	1.16	4.36	186	0.34
22q11.2_dup	sciatica	338588	247	43130	19	31.5	0.63	0.021	0.60	0.37	0.93	194	0.35
22q11.2_dup	coagulation_defects	379172	261	2546	5	1.8	2.82	0.031	2.98	1.12	6.30	248	0.39
22q11.2_dup	irritable_bowel_syndrome	368700	251	13018	15	8.9	1.65	0.045	1.77	1.01	2.87	311	0.45
22q11.2_dup	died_to_2016	370735	252	10983	14	7.5	1.83	0.053	1.77	0.99	2.90	345	0.48
22q11.2_dup	atherosclerotic_vascular_disease	375961	258	5757	8	4.0	2.00	0.071	2.02	0.94	3.78	407	0.54
22q11.2_dup	respiratory	360945	243	20773	23	14.0	1.59	0.076	1.52	0.95	2.29	434	0.55
22q11.2_dup	venous_thromboembolic_disease	367731	251	13987	15	9.5	1.54	0.082	1.63	0.93	2.64	462	0.56
22q11.2_dup	hypertension	260992	171	120726	95	79.1	1.13	0.086	1.26	0.97	1.64	474	0.57
22q11.2_dup	ear	367484	251	14234	15	9.7	1.51	0.097	1.59	0.91	2.57	518	0.59
22q11.2_dup	osteoporosis	370114	255	11604	11	8.0	1.36	0.10	1.73	0.89	3.04	529	0.60
22q11.2_dup	ocular	360038	246	21680	20	14.8	1.32	0.11	1.48	0.91	2.28	541	0.62
22q11.2_dup	obesity	371869	254	9849	12	6.7	1.75	0.12	1.63	0.87	2.76	590	0.63
22q11.2_dup	thyroid	354567	255	27151	11	19.5	0.58	0.12	0.64	0.33	1.11	599	0.63
22q11.2_dup	ovarian_cysts	374322	258	7396	8	5.1	1.55	0.13	1.79	0.82	3.38	640	0.64
22q11.2_dup	neuropathies	365799	250	15919	16	10.9	1.44	0.13	1.50	0.88	2.40	647	0.64
22q11.2_dup	renal_failure	373789	257	7929	9	5.5	1.63	0.14	1.69	0.82	3.08	676	0.67
22q11.2_dup	nasal	378522	262	3196	4	2.2	1.80	0.15	2.17	0.72	4.89	686	0.68
22q11.2_dup	heart_other	369885	254	11833	12	8.1	1.46	0.16	1.55	0.83	2.65	717	0.69
22q11.2_dup	varicose_veins	369862	262	11856	4	8.4	0.48	0.16	0.55	0.18	1.23	726	0.69
22q11.2_dup	osteoarthritis	312959	212	68759	54	46.6	1.13	0.16	1.25	0.91	1.69	734	0.70
22q11.2_dup	diabetes_other	360964	246	20754	20	14.1	1.38	0.21	1.35	0.83	2.09	887	0.76
22q11.2_dup	asthma	332352	224	49366	42	33.3	1.22	0.24	1.22	0.87	1.68	966	0.78
22q11.2_dup	psoriasis	375696	264	6022	2	4.2	0.48	0.30	0.55	0.12	1.56	1173	0.81
22q11.2_dup	migraine	368027	253	13691	13	9.4	1.36	0.31	1.36	0.73	2.31	1201	0.81
22q11.2_dup	COPD	367476	252	14242	14	9.8	1.41	0.34	1.32	0.73	2.19	1283	0.82
22q11.2_dup	aneurism	379750	264	1968	2	1.4	1.46	0.36	1.90	0.40	5.42	1343	0.84
22q11.2_dup	allergy	323013	221	58705	45	40.2	1.10	0.38	1.16	0.83	1.57	1420	0.85
22q11.2_dup	renal	374426	263	7292	3	5.1	0.59	0.42	0.67	0.19	1.64	1510	0.87
22q11.2_dup	ureter_bladder	333154	229	48564	37	33.4	1.09	0.43	1.15	0.80	1.61	1555	0.87
22q11.2_dup	stroke_derived2017	372892	258	8826	8	6.1	1.30	0.44	1.33	0.61	2.49	1574	0.87
22q11.2_dup	gastrointestinal_ulcer	371357	257	10361	9	7.2	1.25	0.48	1.28	0.62	2.32	1696	0.88
22q11.2_dup	biliary	360928	250	20790	16	14.4	1.10	0.48	1.20	0.70	1.92	1718	0.88
22q11.2_dup	congenital	376689	264	5029	2	3.5	0.57	0.49	0.67	0.14	1.88	1743	0.89
22q11.2_dup	cataract	359853	251	21865	15	15.3	0.98	0.51	1.20	0.68	1.96	1788	0.89
22q11.2_dup	uterine_problem	339401	237	42317	29	29.5	0.98	0.63	1.11	0.72	1.65	2083	0.94
22q11.2_dup	any_Ca	331994	238	49724	28	35.6	0.81	0.64	0.91	0.60	1.33	2120	0.94
22q11.2_dup	cerebrovascular_disease_not_stroke	377385	263	4333	3	3.0	0.99	0.68	1.26	0.35	3.12	2231	0.95
22q11.2_dup	heart_valve	373336	260	8382	6	5.8	1.03	0.69	1.18	0.48	2.37	2261	0.95
22q11.2_dup	arrhythmia	357543	249	24175	17	16.8	1.01	0.72	1.10	0.65	1.74	2325	0.96
22q11.2_dup	hepatic	372958	259	8760	7	6.1	1.15	0.76	1.12	0.49	2.16	2465	0.97
22q11.2_dup	diabetes_insulin_dependent	379122	264	2596	2	1.8	1.11	0.80	1.18	0.25	3.34	2587	0.97
22q11.2_dup	inflammatory_bowel_disease	363116	254	18602	12	13.0	0.93	0.80	0.93	0.50	1.57	2588	0.97
22q11.2_dup	diverticular_disease_intestine	354568	249	27150	17	19.1	0.90	0.85	0.95	0.55	1.53	2741	0.97
22q11.2_dup	prostate_hyperplasia	367262	257	14456	9	10.1	0.89	0.90	1.04	0.50	1.95	2876	0.98
22q11.2_dup	paralytic_syndromes	379757	265	1961	1	1.4	0.73	0.93	0.93	0.11	3.34	2957	0.99
22q11.2_dup	anaemia	362565	253	19153	13	13.4	0.97	0.94	0.98	0.54	1.63	2965	0.99
22q11.2_dup	connective_tissue_disorder	372360	260	9358	6	6.5	0.92	0.95	1.03	0.42	2.06	3003	0.99
22q11.2_dup	high_cholesterol	315092	220	66626	46	46.5	0.99	0.96	1.01	0.71	1.40	3044	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
22q11.2distal_del	aneurism	379490	4	1967	1	0.0	38.80	0.0013	104.85	9.71	673.61	46	0.092
22q11.2distal_del	paralytic syndromes	379496	4	1961	1	0.0	38.92	0.0038	51.58	5.07	289.93	71	0.17
22q11.2distal_del	obesity	371618	3	9839	2	0.1	15.51	0.0038	18.77	2.98	100.40	72	0.17
22q11.2distal_del	diabetes insulin dependent	378862	4	2595	1	0.0	29.41	0.0063	37.86	3.70	213.72	88	0.22
22q11.2distal_del	thyroid	354315	3	27142	2	0.2	5.62	0.016	11.34	1.68	68.80	162	0.30
22q11.2distal_del	heart valve	373080	4	8377	1	0.1	9.11	0.018	19.46	1.89	110.43	174	0.33
22q11.2distal_del	renal failure	373536	4	7921	1	0.1	9.63	0.021	18.72	1.75	114.67	189	0.35
22q11.2distal_del	renal	374167	4	7290	1	0.1	10.47	0.023	16.79	1.66	92.62	204	0.35
22q11.2distal_del	stroke derived2017	372638	4	8819	1	0.1	8.65	0.028	15.56	1.46	94.10	233	0.37
22q11.2distal_del	osteoporosis	369863	4	11594	1	0.1	6.58	0.031	16.66	1.37	118.11	247	0.39
22q11.2distal_del	died to 2016	370487	4	10970	1	0.1	6.96	0.052	9.90	0.97	55.97	337	0.49
22q11.2distal_del	ear	367237	4	14220	1	0.2	5.37	0.061	8.92	0.88	49.29	366	0.52
22q11.2distal_del	ocular	359796	4	21661	1	0.2	3.52	0.086	7.14	0.70	40.03	476	0.57
22q11.2distal_del	hypertension	260823	2	120634	3	0.9	1.90	0.096	4.32	0.76	27.80	513	0.59
22q11.2distal_del	diabetes other	360722	4	20735	1	0.2	3.68	0.12	5.94	0.56	35.70	597	0.63
22q11.2distal_del	inflammatory bowel disease	362866	4	18591	1	0.2	4.10	0.12	5.71	0.56	31.61	598	0.63
22q11.2distal_del	osteoarthritis	312750	3	68707	2	0.7	2.22	0.13	4.20	0.63	23.62	630	0.64
22q11.2distal_del	nasal	378265	5	3192	0	0.0	0.00	0.20	14.10	0.11	127.76	825	0.74
22q11.2distal_del	endocrine other	379097	5	2360	0	0.0	0.00	0.20	13.60	0.10	125.43	846	0.74
22q11.2distal_del	low WBC count	378492	5	2965	0	0.0	0.00	0.21	12.44	0.09	111.13	881	0.75
22q11.2distal_del	cerebrovascular disease_not_stroke	377127	5	4330	0	0.1	0.00	0.22	11.53	0.09	112.07	915	0.77
22q11.2distal_del	coagulation defects	378916	5	2541	0	0.0	0.00	0.23	11.02	0.08	99.34	928	0.77
22q11.2distal_del	any_Ca	331760	4	49697	1	0.6	1.54	0.25	3.48	0.34	19.66	988	0.78
22q11.2distal_del	glaucoma	374929	5	6528	0	0.1	0.00	0.27	8.38	0.06	81.24	1073	0.80
22q11.2distal_del	heartfailure	376382	5	5075	0	0.1	0.00	0.30	7.43	0.06	75.31	1146	0.81
22q11.2distal_del	gout	374367	5	7090	0	0.1	0.00	0.33	6.39	0.05	72.35	1253	0.82
22q11.2distal_del	congenital	376430	5	5027	0	0.1	0.00	0.35	5.69	0.04	50.48	1309	0.83
22q11.2distal_del	ureter bladder	332929	4	48528	1	0.6	1.57	0.36	2.60	0.26	14.57	1337	0.84
22q11.2distal_del	digestive	299613	5	81844	0	1.4	0.00	0.37	0.32	0.00	2.90	1366	0.85
22q11.2distal_del	psoriasis	375437	5	6020	0	0.1	0.00	0.38	5.02	0.04	44.39	1389	0.85
22q11.2distal_del	prostate hyperplasia	367010	5	14447	0	0.2	0.00	0.39	5.05	0.04	72.86	1445	0.85
22q11.2distal_del	atherosclerotic vascular disease	375708	5	5749	0	0.1	0.00	0.39	4.79	0.04	48.24	1458	0.85
22q11.2distal_del	heart other	369636	5	11821	0	0.2	0.00	0.48	3.48	0.03	33.21	1695	0.88
22q11.2distal_del	uterine problem	339168	4	42289	1	0.5	1.80	0.49	2.10	0.19	15.87	1730	0.89
22q11.2distal_del	high cholesterol	314876	4	66581	1	0.8	1.15	0.50	2.06	0.19	12.98	1746	0.89
22q11.2distal_del	gastrointestinal ulcer	371105	5	10352	0	0.1	0.00	0.50	3.19	0.02	29.69	1771	0.89
22q11.2distal_del	hepatic	372704	5	8753	0	0.1	0.00	0.50	3.17	0.02	28.68	1773	0.89
22q11.2distal_del	ovarian cysts	374069	5	7388	0	0.1	0.00	0.51	3.11	0.02	32.29	1801	0.89
22q11.2distal_del	connective tissue disorder	372105	5	9352	0	0.1	0.00	0.52	3.07	0.02	30.40	1812	0.90
22q11.2distal_del	MI derived2017	367597	5	13860	0	0.2	0.00	0.53	2.95	0.02	29.86	1845	0.90
22q11.2distal_del	allergy	322796	4	58661	1	0.7	1.30	0.55	1.82	0.18	9.87	1882	0.91
22q11.2distal_del	varicose veins	369605	5	11852	0	0.2	0.00	0.55	2.73	0.02	24.64	1894	0.92
22q11.2distal_del	ischaemic heart disease_not_MI	365134	5	16323	0	0.2	0.00	0.58	2.56	0.02	25.52	1955	0.93
22q11.2distal_del	asthma	332133	5	49324	0	0.7	0.00	0.58	0.48	0.00	4.29	1971	0.93
22q11.2distal_del	cataract	359607	5	21850	0	0.3	0.00	0.61	2.41	0.02	26.87	2036	0.94
22q11.2distal_del	venous thromboembolic disease	367485	5	13972	0	0.2	0.00	0.61	2.35	0.02	21.79	2038	0.94
22q11.2distal_del	migraine	367779	5	13678	0	0.2	0.00	0.64	2.15	0.02	19.91	2132	0.94
22q11.2distal_del	irritable bowel syndrome	368454	5	13003	0	0.2	0.00	0.66	2.03	0.02	18.80	2189	0.95
22q11.2distal_del	arrhythmia	357299	5	24158	0	0.3	0.00	0.72	1.77	0.01	16.74	2355	0.96
22q11.2distal_del	sciatica	338346	5	43111	0	0.6	0.00	0.73	0.62	0.00	5.55	2374	0.96
22q11.2distal_del	neuropathies	365554	5	15903	0	0.2	0.00	0.75	1.66	0.01	15.47	2437	0.97
22q11.2distal_del	diverticular disease_intestine	354324	5	27133	0	0.4	0.00	0.77	1.59	0.01	15.46	2503	0.97
22q11.2distal_del	respiratory	360707	5	20750	0	0.3	0.00	0.82	1.43	0.01	13.21	2637	0.97
22q11.2distal_del	anaemia	362317	5	19140	0	0.3	0.00	0.83	1.41	0.01	12.98	2656	0.97
22q11.2distal_del	biliary	360683	5	20774	0	0.3	0.00	0.83	1.42	0.01	13.58	2660	0.97
22q11.2distal_del	hernia	333569	5	47888	0	0.7	0.00	0.83	0.73	0.01	6.80	2667	0.97
22q11.2distal_del	COPD	367229	5	14228	0	0.2	0.00	0.89	1.25	0.01	15.33	2852	0.98
22q11.2distal_del	gastric reflux	347162	5	34295	0	0.5	0.00	0.93	0.88	0.01	8.04	2958	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
22q11.2distal_dup	paralytic syndromes	379503	11	1961	1	0.1	16.22	0.015	20.10	2.16	85.60	154	0.30
22q11.2distal_dup	stroke_derived2017	372644	10	8820	2	0.2	7.21	0.021	7.74	1.46	27.83	187	0.35
22q11.2distal_dup	arrhythmia	357303	9	24161	3	0.6	3.95	0.032	4.76	1.17	15.35	254	0.39
22q11.2distal_dup	prostate hyperplasia	367015	10	14449	2	0.4	4.40	0.039	7.01	1.12	35.12	283	0.43
22q11.2distal_dup	cerebrovascular disease not stroke	377133	11	4331	1	0.1	7.34	0.046	9.84	1.05	42.62	317	0.46
22q11.2distal_dup	MI_derived2017	367602	10	13862	2	0.4	4.59	0.049	5.49	1.00	20.61	327	0.47
22q11.2distal_dup	heartfailure	376388	11	5076	1	0.1	6.26	0.068	7.81	0.82	34.90	390	0.54
22q11.2distal_dup	high cholesterol	314879	7	66585	5	1.5	2.39	0.078	2.91	0.88	9.03	442	0.55
22q11.2distal_dup	ovarian cysts	374075	11	7389	1	0.2	4.30	0.085	6.93	0.71	34.76	469	0.57
22q11.2distal_dup	glaucoma	374935	11	6529	1	0.2	4.87	0.087	6.54	0.70	28.03	477	0.57
22q11.2distal_dup	atherosclerotic vascular disease	375714	11	5750	1	0.2	5.53	0.089	6.50	0.69	28.55	485	0.58
22q11.2distal_dup	gout	374373	11	7091	1	0.2	4.48	0.097	6.28	0.65	29.93	519	0.59
22q11.2distal_dup	cataract	359612	10	21852	2	0.6	2.91	0.13	3.45	0.65	12.47	635	0.64
22q11.2distal_dup	heart_valve	373087	11	8377	1	0.2	3.80	0.13	4.94	0.53	20.98	643	0.64
22q11.2distal_dup	heart_other	369642	11	11822	1	0.4	2.69	0.23	3.44	0.37	14.80	920	0.77
22q11.2distal_dup	osteoporosis	369870	11	11594	1	0.3	2.74	0.24	3.41	0.35	15.95	951	0.78
22q11.2distal_dup	ear	367244	11	14220	1	0.4	2.24	0.29	2.86	0.31	12.06	1127	0.81
22q11.2distal_dup	uterine problem	339176	12	42288	0	1.5	0.00	0.31	0.29	0.00	2.41	1179	0.81
22q11.2distal_dup	aneurism	379498	12	1966	0	0.1	0.00	0.32	6.26	0.05	49.86	1237	0.82
22q11.2distal_dup	osteoarthritis	312758	11	68706	1	2.4	0.46	0.33	0.46	0.05	1.97	1246	0.82
22q11.2distal_dup	hypertension	260827	6	120637	6	2.8	1.58	0.35	1.70	0.55	5.32	1318	0.83
22q11.2distal_dup	endocrine_other	379104	12	2360	0	0.1	0.00	0.35	5.46	0.04	42.24	1322	0.83
22q11.2distal_dup	coagulation_defects	378923	12	2541	0	0.1	0.00	0.36	5.22	0.04	39.79	1344	0.84
22q11.2distal_dup	low_WBC_count	378499	12	2965	0	0.1	0.00	0.38	4.90	0.04	37.36	1394	0.85
22q11.2distal_dup	ischaemic heart disease not MI	365140	11	16324	1	0.5	1.95	0.38	2.34	0.25	10.23	1418	0.85
22q11.2distal_dup	diabetes insulin dependent	378870	12	2594	0	0.1	0.00	0.39	4.65	0.04	35.76	1441	0.85
22q11.2distal_dup	nasal	378272	12	3192	0	0.1	0.00	0.40	4.56	0.04	35.02	1461	0.85
22q11.2distal_dup	gastric reflux	347169	12	34295	0	1.2	0.00	0.40	0.36	0.00	2.73	1476	0.85
22q11.2distal_dup	anaemia	362323	11	19141	1	0.6	1.66	0.46	2.03	0.22	8.73	1642	0.88
22q11.2distal_dup	COPD	367235	11	14229	1	0.4	2.23	0.47	2.09	0.20	10.26	1670	0.88
22q11.2distal_dup	ocular	359803	11	21661	1	0.7	1.47	0.49	1.92	0.21	8.09	1732	0.89
22q11.2distal_dup	thyroid	354324	12	27140	0	0.9	0.00	0.51	0.43	0.00	3.46	1786	0.90
22q11.2distal_dup	diverticular disease intestine	354331	12	27133	0	0.9	0.00	0.51	0.43	0.00	3.37	1789	0.89
22q11.2distal_dup	congenital	376437	12	5027	0	0.2	0.00	0.54	2.78	0.02	21.16	1864	0.91
22q11.2distal_dup	asthma	332138	10	49326	2	1.5	1.29	0.54	1.57	0.30	5.48	1867	0.91
22q11.2distal_dup	any_Ca	331766	10	49698	2	1.5	1.28	0.61	1.45	0.28	5.08	2042	0.94
22q11.2distal_dup	psoriasis	375444	12	6020	0	0.2	0.00	0.61	2.28	0.02	17.39	2048	0.94
22q11.2distal_dup	diabetes_other	360730	12	20734	0	0.7	0.00	0.63	0.53	0.00	4.16	2108	0.94
22q11.2distal_dup	ureter_bladder	332935	10	48529	2	1.5	1.31	0.64	1.41	0.27	4.92	2119	0.94
22q11.2distal_dup	respiratory	360714	12	20750	0	0.7	0.00	0.66	0.56	0.00	4.29	2168	0.95
22q11.2distal_dup	biliary	360690	12	20774	0	0.7	0.00	0.68	0.58	0.00	4.52	2237	0.95
22q11.2distal_dup	allergy	322803	11	58661	1	2.0	0.54	0.72	0.74	0.08	3.11	2327	0.96
22q11.2distal_dup	renal	374175	12	7289	0	0.2	0.00	0.72	1.76	0.01	13.44	2340	0.96
22q11.2distal_dup	hernia	333575	11	47889	1	1.6	0.66	0.75	0.76	0.08	3.25	2428	0.97
22q11.2distal_dup	inflammatory bowel disease	362874	12	18590	0	0.6	0.00	0.76	0.66	0.01	5.03	2449	0.97
22q11.2distal_dup	renal failure	373544	12	7920	0	0.3	0.00	0.82	1.42	0.01	11.13	2635	0.97
22q11.2distal_dup	hepatic	372711	12	8753	0	0.3	0.00	0.83	1.39	0.01	10.74	2666	0.97
22q11.2distal_dup	digestive	299617	9	81847	3	2.5	1.17	0.84	1.14	0.28	3.64	2696	0.97
22q11.2distal_dup	neuropathies	365561	12	15903	0	0.5	0.00	0.84	0.76	0.01	5.85	2711	0.97
22q11.2distal_dup	connective_tissue_disorder	372112	12	9352	0	0.3	0.00	0.89	1.22	0.01	9.68	2855	0.98
22q11.2distal_dup	sciatica	338352	11	43112	1	1.4	0.74	0.90	0.90	0.10	3.78	2872	0.98
22q11.2distal_dup	venous thromboembolic disease	367492	12	13972	0	0.5	0.00	0.92	0.86	0.01	6.60	2913	0.98
22q11.2distal_dup	gastrointestinal ulcer	371112	12	10352	0	0.3	0.00	0.93	1.14	0.01	8.93	2954	0.98
22q11.2distal_dup	migraine	367786	12	13678	0	0.4	0.00	0.94	1.13	0.01	8.80	2967	0.99
22q11.2distal_dup	varicose veins	369612	12	11852	0	0.4	0.00	0.94	1.12	0.01	8.59	2972	0.99
22q11.2distal_dup	died to 2016	370495	12	10969	0	0.4	0.00	0.94	1.11	0.01	8.61	2987	0.99
22q11.2distal_dup	irritable_bowel_syndrome	368461	12	13003	0	0.4	0.00	0.95	1.10	0.01	8.56	3005	0.99
22q11.2distal_dup	obesity	371627	12	9837	0	0.3	0.00	0.96	1.07	0.01	8.51	3046	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q11.2 del	obesity	371641	26	9840	3	0.7	4.01	0.029	4.48	1.20	12.20	241	0.38
2q11.2 del	osteoarthritis	312768	21	68713	8	4.6	1.53	0.078	2.19	0.91	4.82	441	0.56
2q11.2 del	nasal	378288	28	3193	1	0.2	4.12	0.080	6.62	0.74	25.36	452	0.56
2q11.2 del	cerebrovascular_disease_not_stroke	377150	28	4331	1	0.3	3.04	0.13	5.00	0.55	19.65	618	0.63
2q11.2 del	ocular	359818	26	21663	3	1.6	1.82	0.16	2.46	0.66	6.67	724	0.70
2q11.2 del	irritable_bowel_syndrome	368476	27	13005	2	1.0	2.02	0.18	2.74	0.55	8.45	791	0.73
2q11.2 del	glaucoma	374952	28	6529	1	0.5	2.01	0.20	3.69	0.41	14.46	829	0.74
2q11.2 del	diverticular_disease_intestine	354348	29	27133	0	2.2	0.00	0.21	0.24	0.00	1.77	876	0.75
2q11.2 del	ear	367260	27	14221	2	1.0	1.85	0.25	2.38	0.48	7.26	982	0.78
2q11.2 del	hypertension	260844	23	120637	6	10.6	0.65	0.26	0.61	0.23	1.42	1026	0.79
2q11.2 del	neuropathies	365576	27	15905	2	1.2	1.65	0.28	2.22	0.45	6.78	1094	0.80
2q11.2 del	gastric_reflux	347182	25	34299	4	2.5	1.53	0.28	1.81	0.57	4.51	1106	0.80
2q11.2 del	inflammatory_bowel_disease	362891	29	18590	0	1.5	0.00	0.33	0.32	0.00	2.29	1265	0.82
2q11.2 del	cataract	359629	27	21852	2	1.6	1.20	0.34	2.04	0.40	6.59	1284	0.82
2q11.2 del	connective_tissue_disorder	372128	28	9353	1	0.7	1.41	0.36	2.39	0.27	9.22	1341	0.84
2q11.2 del	uterine_problem	339192	28	42289	1	3.5	0.31	0.38	0.50	0.05	2.09	1416	0.85
2q11.2 del	hernia	333588	24	47893	5	3.4	1.37	0.39	1.54	0.54	3.71	1439	0.85
2q11.2 del	digestive	299629	21	81852	8	5.7	1.29	0.40	1.43	0.60	3.09	1471	0.85
2q11.2 del	biliary	360705	27	20776	2	1.6	1.27	0.42	1.78	0.36	5.48	1526	0.87
2q11.2 del	ischaemic_heart_disease_not_MI	365158	29	16323	0	1.3	0.00	0.42	0.38	0.00	2.76	1530	0.87
2q11.2 del	prostate_hyperplasia	367034	29	14447	0	1.1	0.00	0.44	0.38	0.00	2.97	1562	0.87
2q11.2 del	anaemia	362339	27	19142	2	1.4	1.37	0.45	1.72	0.35	5.26	1606	0.87
2q11.2 del	high_cholesterol	314895	23	66586	6	4.9	1.19	0.47	1.42	0.52	3.38	1666	0.88
2q11.2 del	aneurism	379515	29	1966	0	0.2	0.00	0.48	3.39	0.03	25.09	1691	0.88
2q11.2 del	hepatic	372727	28	8754	1	0.7	1.50	0.48	1.93	0.22	7.40	1692	0.88
2q11.2 del	endocrine_other	379121	29	2360	0	0.2	0.00	0.53	2.85	0.02	20.33	1840	0.90
2q11.2 del	venous_thromboembolic_disease	367509	29	13972	0	1.1	0.00	0.54	0.46	0.00	3.30	1851	0.91
2q11.2 del	died_to_2016	370512	29	10969	0	0.9	0.00	0.55	0.47	0.00	3.40	1892	0.92
2q11.2 del	paralytic_syndromes	379521	29	1960	0	0.1	0.00	0.56	2.63	0.02	18.95	1909	0.91
2q11.2 del	migraine	367803	29	13678	0	1.1	0.00	0.58	0.50	0.00	3.58	1966	0.93
2q11.2 del	coagulation_defects	378940	29	2541	0	0.2	0.00	0.59	2.42	0.02	17.21	1983	0.93
2q11.2 del	any_Ca	331783	27	49698	2	4.0	0.53	0.59	0.71	0.14	2.19	1992	0.93
2q11.2 del	gastrointestinal_ulcer	371128	28	10353	1	0.8	1.27	0.60	1.62	0.18	6.29	2001	0.93
2q11.2 del	heart_other	369660	29	11821	0	0.9	0.00	0.60	0.51	0.00	3.72	2013	0.94
2q11.2 del	low_WBC_count	378516	29	2965	0	0.2	0.00	0.61	2.30	0.02	16.38	2025	0.94
2q11.2 del	respiratory	360729	27	20752	2	1.6	1.27	0.62	1.42	0.29	4.37	2058	0.94
2q11.2 del	arrhythmia	357321	27	24160	2	1.8	1.09	0.63	1.40	0.28	4.38	2094	0.94
2q11.2 del	varicose_veins	369629	29	11852	0	0.9	0.00	0.65	0.55	0.00	3.93	2139	0.95
2q11.2 del	diabetes_insulin_dependent	378887	29	2594	0	0.2	0.00	0.65	2.07	0.02	14.90	2147	0.95
2q11.2 del	stroke_derived2017	372663	29	8818	0	0.7	0.00	0.76	0.67	0.01	4.83	2467	0.97
2q11.2 del	asthma	332154	26	49327	3	3.9	0.80	0.78	0.86	0.23	2.30	2528	0.97
2q11.2 del	gout	374391	29	7090	0	0.5	0.00	0.81	0.72	0.01	5.31	2608	0.97
2q11.2 del	MI_derived2017	367620	28	13861	1	1.1	0.95	0.81	1.24	0.14	4.96	2610	0.97
2q11.2 del	diabetes_other	360746	28	20735	1	1.6	0.63	0.83	0.84	0.09	3.28	2677	0.97
2q11.2 del	COPD	367252	28	14229	1	1.1	0.92	0.84	1.20	0.13	4.76	2689	0.97
2q11.2 del	osteoporosis	369888	29	11593	0	0.9	0.00	0.85	0.77	0.01	5.78	2743	0.97
2q11.2 del	renal_failure	373561	29	7920	0	0.6	0.00	0.86	0.78	0.01	5.66	2756	0.97
2q11.2 del	thyroid	354340	28	27141	1	2.1	0.48	0.86	0.86	0.09	3.38	2757	0.97
2q11.2 del	heart_valve	373105	29	8376	0	0.7	0.00	0.88	0.82	0.01	5.86	2821	0.98
2q11.2 del	ureter_bladder	332951	26	48530	3	3.8	0.81	0.89	0.92	0.25	2.50	2828	0.98
2q11.2 del	renal	374192	29	7289	0	0.6	0.00	0.89	0.83	0.01	5.92	2860	0.98
2q11.2 del	congenital	376454	29	5027	0	0.4	0.00	0.92	1.16	0.01	8.21	2930	0.98
2q11.2 del	allergy	322817	25	58664	4	4.5	0.90	0.94	1.04	0.33	2.58	2975	0.99
2q11.2 del	heartfailure	376406	29	5075	0	0.4	0.00	0.94	1.12	0.01	8.26	2980	0.99
2q11.2 del	psoriasis	375461	29	6020	0	0.5	0.00	0.96	0.93	0.01	6.63	3036	0.99
2q11.2 del	sciatica	338367	26	43114	3	3.3	0.92	0.97	1.02	0.27	2.75	3062	0.99
2q11.2 del	atherosclerotic_vascular_disease	375732	29	5749	0	0.4	0.00	0.99	1.01	0.01	7.33	3118	1.00
2q11.2 del	ovarian_cysts	374093	29	7388	0	0.6	0.00	1.00	1.01	0.01	7.67	3122	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q11.2_dup	inflammatory_bowel_disease	362883	21	18595	5	1.1	3.95	0.0040	5.16	1.81	12.37	73	0.17
2q11.2_dup	cerebrovascular_disease_not_stroke	377146	24	4332	2	0.3	6.78	0.0088	10.31	2.05	32.80	110	0.25
2q11.2_dup	ovarian_cysts	374088	24	7390	2	0.5	3.97	0.042	5.54	1.08	18.61	299	0.44
2q11.2_dup	ischaemic_heart_disease_not_MI	365152	23	16326	3	1.0	2.70	0.043	4.05	1.05	11.48	300	0.45
2q11.2_dup	hepatic	372723	24	8755	2	0.6	3.35	0.061	4.57	0.92	14.12	365	0.52
2q11.2_dup	cataract	359625	23	21853	3	1.4	2.01	0.11	2.92	0.75	8.52	557	0.62
2q11.2_dup	ocular	359815	23	21663	3	1.4	2.03	0.13	2.67	0.71	7.34	641	0.64
2q11.2_dup	ear	367257	24	14221	2	0.9	2.06	0.17	2.83	0.57	8.74	755	0.72
2q11.2_dup	psoriasis	375457	25	6021	1	0.4	2.44	0.19	3.74	0.42	14.36	811	0.74
2q11.2_dup	gout	374387	25	7091	1	0.5	2.07	0.20	3.71	0.40	15.18	835	0.75
2q11.2_dup	ureter_bladder	332946	21	48532	5	3.1	1.51	0.22	1.88	0.66	4.54	900	0.76
2q11.2_dup	any_Ca	331777	21	49701	5	3.1	1.48	0.23	1.85	0.64	4.52	943	0.78
2q11.2_dup	diverticular_disease_intestine	354342	23	27136	3	1.8	1.62	0.24	2.12	0.56	5.98	964	0.78
2q11.2_dup	allergy	322812	20	58666	6	3.6	1.50	0.24	1.75	0.66	4.02	965	0.78
2q11.2_dup	osteoarthritis	312771	24	68707	2	5.3	0.43	0.24	0.49	0.10	1.54	981	0.78
2q11.2_dup	renal	374188	25	7290	1	0.5	2.01	0.25	3.12	0.35	11.97	1001	0.78
2q11.2_dup	heart_valve	373101	25	8377	1	0.6	1.75	0.28	2.84	0.32	11.04	1115	0.80
2q11.2_dup	asthma	332149	21	49329	5	3.1	1.49	0.30	1.69	0.59	4.04	1159	0.81
2q11.2_dup	arrhythmia	357320	26	24158	0	1.8	0.00	0.31	0.30	0.00	2.19	1177	0.81
2q11.2_dup	diabetes_other	360742	24	20736	2	1.4	1.42	0.32	2.09	0.42	6.56	1227	0.81
2q11.2_dup	high_cholesterol	314896	24	66582	2	5.1	0.44	0.32	0.53	0.11	1.71	1230	0.82
2q11.2_dup	biliary	360704	26	20774	0	1.5	0.00	0.35	0.33	0.00	2.37	1307	0.83
2q11.2_dup	anaemia	362336	24	19142	2	1.3	1.53	0.35	1.98	0.40	6.14	1310	0.83
2q11.2_dup	sciatica	338363	22	43115	4	2.8	1.36	0.35	1.67	0.53	4.22	1312	0.83
2q11.2_dup	obesity	371640	25	9838	1	0.7	1.49	0.37	2.34	0.26	9.04	1371	0.85
2q11.2_dup	respiratory	360726	24	20752	2	1.4	1.41	0.38	1.91	0.38	5.91	1388	0.85
2q11.2_dup	gastrointestinal_ulcer	371125	25	10353	1	0.7	1.42	0.39	2.26	0.25	8.82	1446	0.85
2q11.2_dup	aneurism	379512	26	1966	0	0.1	0.00	0.40	4.45	0.03	32.83	1474	0.85
2q11.2_dup	died_to_2016	370508	25	10970	1	0.7	1.34	0.43	2.10	0.23	8.14	1538	0.87
2q11.2_dup	paralytic_syndromes	379518	26	1960	0	0.1	0.00	0.44	3.89	0.03	27.98	1563	0.87
2q11.2_dup	prostate_hyperplasia	367030	25	14448	1	1.0	1.02	0.49	1.97	0.20	9.50	1731	0.89
2q11.2_dup	endocrine_other	379118	26	2360	0	0.2	0.00	0.50	3.08	0.02	22.14	1774	0.89
2q11.2_dup	diabetes_insulin_dependent	378884	26	2594	0	0.2	0.00	0.52	2.95	0.02	21.21	1811	0.90
2q11.2_dup	hypertension	260841	20	120637	6	9.2	0.73	0.52	0.75	0.28	1.77	1820	0.90
2q11.2_dup	coagulation_defects	378937	26	2541	0	0.2	0.00	0.52	2.92	0.02	20.85	1823	0.90
2q11.2_dup	neuropathies	365575	26	15903	0	1.1	0.00	0.53	0.45	0.00	3.25	1835	0.90
2q11.2_dup	irritable_bowel_syndrome	368474	25	13004	1	0.9	1.13	0.56	1.70	0.19	6.58	1916	0.91
2q11.2_dup	COPD	367249	25	14229	1	1.0	1.03	0.57	1.70	0.19	6.87	1932	0.92
2q11.2_dup	low_WBC_count	378513	26	2965	0	0.2	0.00	0.59	2.40	0.02	17.16	1989	0.93
2q11.2_dup	migraine	367800	26	13678	0	1.0	0.00	0.60	0.51	0.00	3.67	2002	0.93
2q11.2_dup	nasal	378286	26	3192	0	0.2	0.00	0.61	2.28	0.02	16.41	2040	0.94
2q11.2_dup	venous_thromboembolic_disease	367506	26	13972	0	1.0	0.00	0.61	0.52	0.00	3.74	2044	0.94
2q11.2_dup	osteoporosis	369885	26	11593	0	0.8	0.00	0.65	0.55	0.00	4.19	2149	0.94
2q11.2_dup	thyroid	354336	24	27142	2	1.8	1.08	0.69	1.32	0.26	4.24	2275	0.95
2q11.2_dup	varicose_veins	369626	26	11852	0	0.8	0.00	0.70	0.60	0.00	4.32	2292	0.96
2q11.2_dup	MI_derived2017	367618	26	13860	0	1.0	0.00	0.72	0.62	0.00	4.57	2328	0.96
2q11.2_dup	heartfailure	376403	26	5075	0	0.4	0.00	0.73	1.72	0.01	12.66	2371	0.96
2q11.2_dup	heart_other	369657	26	11821	0	0.8	0.00	0.76	0.66	0.01	4.78	2451	0.97
2q11.2_dup	uterine_problem	339187	23	42291	3	2.9	1.04	0.76	1.21	0.31	3.66	2458	0.96
2q11.2_dup	atherosclerotic_vascular_disease	375729	26	5749	0	0.4	0.00	0.82	1.41	0.01	10.23	2647	0.97
2q11.2_dup	congenital	376451	26	5027	0	0.3	0.00	0.83	1.39	0.01	9.92	2665	0.97
2q11.2_dup	hernia	333587	23	47891	3	3.3	0.92	0.83	1.14	0.30	3.15	2668	0.97
2q11.2_dup	connective_tissue_disorder	372126	26	9352	0	0.7	0.00	0.85	0.78	0.01	5.65	2751	0.97
2q11.2_dup	gastric_reflux	347181	24	34297	2	2.4	0.86	0.89	1.10	0.22	3.39	2856	0.98
2q11.2_dup	glaucoma	374950	26	6528	0	0.5	0.00	0.91	1.19	0.01	8.72	2886	0.98
2q11.2_dup	stroke_derived2017	372660	26	8818	0	0.6	0.00	0.95	0.92	0.01	6.63	3011	0.99
2q11.2_dup	digestive	299629	21	81849	5	5.7	0.90	0.97	0.98	0.34	2.37	3065	0.99
2q11.2_dup	renal_failure	373558	26	7920	0	0.6	0.00	0.98	1.04	0.01	7.55	3084	0.99



CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q13_del	ureter_bladder	332962	37	48541	14	5.4	2.16	0.0018	2.91	1.52	5.27	53	0.11
2q13_del	connective_tissue_disorder	372146	46	9357	5	1.2	4.00	0.0022	5.58	2.02	12.58	58	0.12
2q13_del	biliary	360722	44	20781	7	2.5	2.52	0.0077	3.41	1.44	7.05	102	0.24
2q13_del	varicose_veins	369646	46	11857	5	1.5	3.16	0.010	3.98	1.45	8.88	125	0.26
2q13_del	cataract	359647	45	21856	6	2.7	2.05	0.015	3.41	1.31	7.61	155	0.30
2q13_del	coagulation_defects	378960	49	2543	2	0.3	5.89	0.019	7.24	1.49	21.27	175	0.34
2q13_del	low_WBC_count	378536	49	2967	2	0.4	5.05	0.021	7.00	1.44	20.54	188	0.35
2q13_del	diverticular_disease_intestine	354363	44	27140	7	3.4	1.93	0.026	2.73	1.14	5.71	224	0.37
2q13_del	asthma	332167	39	49336	12	5.8	1.82	0.040	2.04	1.04	3.76	290	0.43
2q13_del	renal	374211	48	7292	3	0.9	3.08	0.047	3.72	1.02	9.60	319	0.46
2q13_del	hypertension	260851	30	120652	21	13.9	1.30	0.054	1.79	0.99	3.20	349	0.49
2q13_del	prostate_hyperplasia	367052	47	14451	4	1.9	2.07	0.087	2.80	0.84	7.52	480	0.57
2q13_del	aneurism	379536	50	1967	1	0.3	3.80	0.088	6.22	0.69	23.71	482	0.57
2q13_del	hernia	333605	41	47898	10	5.9	1.56	0.10	1.85	0.88	3.57	531	0.60
2q13_del	endocrine_other	379142	50	2361	1	0.3	3.17	0.12	5.13	0.58	19.01	581	0.63
2q13_del	paralytic_syndromes	379542	50	1961	1	0.3	3.82	0.12	5.01	0.56	18.63	606	0.63
2q13_del	COPD	367271	47	14232	4	1.8	2.10	0.13	2.38	0.75	5.87	627	0.64
2q13_del	ovarian_cysts	374113	49	7390	2	1.0	2.02	0.16	2.99	0.60	9.36	706	0.69
2q13_del	nasal	378310	50	3193	1	0.4	2.34	0.19	3.68	0.42	13.62	815	0.74
2q13_del	MI_derived2017	367643	51	13860	0	1.9	0.00	0.20	0.24	0.00	1.72	842	0.74
2q13_del	osteoporosis	369908	49	11595	2	1.5	1.29	0.24	2.40	0.48	7.36	974	0.78
2q13_del	venous_thromboembolic_disease	367528	48	13975	3	1.8	1.61	0.25	2.02	0.55	5.24	1004	0.78
2q13_del	anaemia	362359	47	19144	4	2.5	1.56	0.26	1.84	0.60	4.36	1023	0.79
2q13_del	cerebrovascular_disease_not_stroke	377172	50	4331	1	0.6	1.73	0.26	2.99	0.34	11.25	1033	0.79
2q13_del	hepatic	372748	49	8755	2	1.2	1.71	0.34	1.99	0.41	5.85	1287	0.82
2q13_del	respiratory	360749	47	20754	4	2.7	1.44	0.38	1.60	0.52	3.81	1396	0.85
2q13_del	digestive	299651	43	81852	8	11.7	0.73	0.40	0.73	0.33	1.46	1468	0.85
2q13_del	thyroid	354359	47	27144	4	3.6	1.10	0.41	1.56	0.50	3.79	1490	0.86
2q13_del	heart_other	369680	49	11823	2	1.6	1.27	0.47	1.66	0.34	4.96	1669	0.88
2q13_del	stroke_derived2017	372685	51	8818	0	1.2	0.00	0.48	0.42	0.00	2.94	1689	0.88
2q13_del	atherosclerotic_vascular_disease	375753	50	5750	1	0.8	1.30	0.49	1.88	0.21	7.06	1733	0.89
2q13_del	sciatica	338385	44	43118	7	5.6	1.21	0.50	1.32	0.56	2.70	1758	0.89
2q13_del	gout	374413	51	7090	0	1.0	0.00	0.53	0.46	0.00	3.25	1841	0.90
2q13_del	inflammatory_bowel_disease	362910	48	18593	3	2.5	1.21	0.55	1.42	0.39	3.65	1888	0.91
2q13_del	psoriasis	375482	50	6021	1	0.8	1.24	0.56	1.70	0.19	6.27	1901	0.92
2q13_del	allergy	322834	42	58669	9	7.6	1.15	0.56	1.24	0.58	2.41	1904	0.91
2q13_del	any_Ca	331803	47	49700	4	7.0	0.60	0.59	0.77	0.25	1.84	1975	0.93
2q13_del	migraine	367823	49	13680	2	1.8	1.09	0.61	1.42	0.29	4.20	2046	0.94
2q13_del	heart_valve	373126	50	8377	1	1.1	0.89	0.66	1.47	0.17	5.48	2178	0.95
2q13_del	renal_failure	373582	50	7921	1	1.1	0.94	0.67	1.46	0.16	5.50	2195	0.95
2q13_del	ischaemic_heart_disease_not_MI	365178	49	16325	2	2.2	0.92	0.75	1.24	0.25	3.72	2448	0.97
2q13_del	glaucoma	374975	51	6528	0	0.9	0.00	0.76	0.66	0.01	4.68	2457	0.97
2q13_del	ear	367283	50	14220	1	1.9	0.53	0.77	0.79	0.09	2.92	2487	0.97
2q13_del	congenital	376476	51	5027	0	0.7	0.00	0.77	0.68	0.01	4.74	2507	0.97
2q13_del	arrhythmia	357342	48	24161	3	3.2	0.93	0.78	1.18	0.32	3.08	2516	0.97
2q13_del	osteoarthritis	312791	44	68712	7	9.7	0.76	0.79	0.90	0.37	1.88	2550	0.97
2q13_del	neuropathies	365598	49	15905	2	2.1	0.94	0.79	1.20	0.25	3.52	2552	0.97
2q13_del	diabetes_other	360767	49	20736	2	2.8	0.72	0.79	0.84	0.17	2.53	2564	0.97
2q13_del	heartfailure	376428	51	5075	0	0.7	0.00	0.79	0.70	0.01	5.01	2566	0.97
2q13_del	uterine_problem	339211	47	42292	4	5.9	0.71	0.83	0.89	0.28	2.30	2670	0.97
2q13_del	irritable_bowel_syndrome	368499	50	13004	1	1.8	0.58	0.87	0.88	0.10	3.25	2789	0.98
2q13_del	died_to_2016	370533	50	10970	1	1.5	0.68	0.89	0.89	0.10	3.32	2841	0.98
2q13_del	diabetes_insulin_dependent	378909	51	2594	0	0.3	0.00	0.89	1.21	0.01	8.50	2861	0.98
2q13_del	ocular	359841	49	21662	2	2.9	0.69	0.93	0.94	0.19	2.78	2944	0.99
2q13_del	gastrointestinal_ulcer	371150	50	10353	1	1.4	0.72	0.97	1.03	0.12	3.85	3063	0.99
2q13_del	high_cholesterol	314915	43	66588	8	9.1	0.90	0.97	1.01	0.44	2.09	3071	0.99
2q13_del	gastric_reflux	347204	47	34299	4	4.6	0.87	0.99	0.99	0.32	2.35	3097	1.00
2q13_del	obesity	371665	50	9838	1	1.3	0.76	0.99	1.01	0.11	3.76	3100	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q13_dup	diabetes_other	360778	60	20745	11	3.5	2.85	0.0012	3.39	1.68	6.29	41	0.094
2q13_dup	sciatica	338410	69	43113	2	8.8	0.25	0.017	0.29	0.06	0.83	169	0.32
2q13_dup	ocular	359863	71	21660	0	4.3	0.00	0.026	0.12	0.00	0.83	222	0.36
2q13_dup	hernia	333631	67	47892	4	9.6	0.45	0.070	0.45	0.15	1.06	404	0.54
2q13_dup	psoriasis	375500	68	6023	3	1.1	2.68	0.077	3.12	0.86	7.91	437	0.55
2q13_dup	ischaemic_heart_disease_not_MI	365194	65	16329	6	2.9	1.97	0.078	2.28	0.90	4.86	440	0.55
2q13_dup	renal_failure	373600	68	7923	3	1.4	2.04	0.16	2.42	0.66	6.25	719	0.69
2q13_dup	digestive	299669	61	81854	10	16.7	0.66	0.16	0.64	0.31	1.18	722	0.69
2q13_dup	asthma	332186	58	49337	13	8.6	1.42	0.18	1.52	0.81	2.67	795	0.73
2q13_dup	biliary	360748	70	20775	1	4.0	0.26	0.18	0.39	0.04	1.44	796	0.73
2q13_dup	endocrine_other	379162	70	2361	1	0.4	2.28	0.20	3.54	0.40	12.97	854	0.74
2q13_dup	obesity	371686	71	9837	0	1.9	0.00	0.23	0.26	0.00	1.81	926	0.77
2q13_dup	coagulation_defects	378981	70	2542	1	0.5	2.11	0.24	3.16	0.36	11.54	962	0.78
2q13_dup	osteoporosis	369930	71	11593	0	2.2	0.00	0.25	0.27	0.00	1.91	996	0.78
2q13_dup	arrhythmia	357363	69	24160	2	4.7	0.44	0.27	0.52	0.11	1.53	1055	0.80
2q13_dup	ear	367300	67	14223	4	2.6	1.51	0.28	1.76	0.58	4.11	1111	0.80
2q13_dup	stroke_derived2017	372705	71	8818	0	1.7	0.00	0.28	0.29	0.00	2.05	1112	0.80
2q13_dup	inflammatory_bowel_disease	362928	66	18595	5	3.4	1.45	0.29	1.66	0.61	3.62	1123	0.81
2q13_dup	gastrointestinal_ulcer	371168	68	10355	3	1.9	1.56	0.30	1.86	0.51	4.77	1170	0.81
2q13_dup	thyroid	354377	65	27146	6	5.0	1.19	0.37	1.48	0.59	3.14	1384	0.85
2q13_dup	gastric_reflux	347220	63	34303	8	6.2	1.25	0.38	1.40	0.63	2.72	1401	0.85
2q13_dup	ovarian_cysts	374133	69	7390	2	1.4	1.45	0.38	1.88	0.38	5.63	1402	0.85
2q13_dup	varicose_veins	369668	68	11855	3	2.2	1.36	0.39	1.67	0.46	4.24	1429	0.85
2q13_dup	uterine_problem	339230	66	42293	5	8.2	0.64	0.39	0.68	0.25	1.57	1442	0.85
2q13_dup	renal	374234	71	7289	0	1.4	0.00	0.40	0.37	0.00	2.52	1464	0.85
2q13_dup	COPD	367294	70	14229	1	2.7	0.38	0.40	0.53	0.06	1.98	1469	0.85
2q13_dup	respiratory	360771	69	20752	2	4.0	0.52	0.42	0.62	0.13	1.79	1520	0.87
2q13_dup	allergy	322850	58	58673	13	10.5	1.19	0.44	1.27	0.67	2.23	1572	0.87
2q13_dup	prostate_hyperplasia	367072	67	14451	4	2.6	1.49	0.45	1.50	0.47	3.80	1619	0.88
2q13_dup	venous_thromboembolic_disease	367550	70	13973	1	2.7	0.38	0.47	0.58	0.07	2.11	1656	0.88
2q13_dup	glaucoma	374995	71	6528	0	1.2	0.00	0.47	0.42	0.00	2.89	1674	0.88
2q13_dup	cataract	359671	69	21852	2	4.2	0.49	0.47	0.64	0.13	1.91	1676	0.88
2q13_dup	diverticular_disease_intestine	354387	68	27136	3	5.2	0.59	0.51	0.71	0.19	1.81	1784	0.89
2q13_dup	atherosclerotic_vascular_disease	375774	71	5749	0	1.1	0.00	0.52	0.45	0.00	3.14	1813	0.90
2q13_dup	connective_tissue_disorder	372169	69	9354	2	1.7	1.15	0.52	1.56	0.32	4.55	1816	0.90
2q13_dup	high_cholesterol	314929	57	66594	14	12.1	1.13	0.53	1.22	0.64	2.19	1837	0.90
2q13_dup	irritable_bowel_syndrome	368519	70	13004	1	2.5	0.41	0.53	0.62	0.07	2.27	1846	0.90
2q13_dup	hepatic	372768	69	8755	2	1.6	1.23	0.54	1.52	0.32	4.41	1860	0.91
2q13_dup	hypertension	260868	47	120655	24	21.7	1.07	0.56	1.17	0.69	1.94	1918	0.91
2q13_dup	anaemia	362381	69	19142	2	3.6	0.56	0.56	0.70	0.15	2.03	1921	0.92
2q13_dup	migraine	367842	68	13681	3	2.5	1.18	0.58	1.38	0.38	3.53	1951	0.93
2q13_dup	any_Ca	331820	64	49703	7	9.6	0.76	0.59	0.81	0.35	1.64	1987	0.93
2q13_dup	congenital	376496	71	5027	0	0.9	0.00	0.60	0.51	0.00	3.54	2008	0.93
2q13_dup	heartfailure	376447	70	5076	1	0.9	1.06	0.65	1.50	0.17	5.59	2145	0.95
2q13_dup	MI_derived2017	367660	68	13863	3	2.6	1.16	0.68	1.27	0.35	3.33	2229	0.95
2q13_dup	cerebrovascular_disease_not_stroke	377193	71	4330	0	0.8	0.00	0.72	0.63	0.00	4.37	2362	0.96
2q13_dup	neuropathies	365617	68	15906	3	3.0	1.01	0.72	1.22	0.34	3.11	2369	0.96
2q13_dup	died_to_2016	370552	69	10971	2	2.0	0.98	0.81	1.17	0.24	3.40	2624	0.97
2q13_dup	paralytic_syndromes	379563	71	1960	0	0.4	0.00	0.85	1.33	0.01	9.19	2733	0.97
2q13_dup	ureter_bladder	332987	62	48536	9	9.0	1.00	0.85	1.07	0.50	2.03	2736	0.97
2q13_dup	aneurism	379557	71	1966	0	0.4	0.00	0.87	1.27	0.01	8.89	2794	0.98
2q13_dup	nasal	378331	71	3192	0	0.6	0.00	0.88	0.81	0.01	5.60	2811	0.98
2q13_dup	heart_other	369700	69	11823	2	2.2	0.91	0.89	1.10	0.23	3.21	2837	0.98
2q13_dup	osteoarthritis	312806	59	68717	12	13.0	0.94	0.92	1.03	0.53	1.87	2917	0.98
2q13_dup	low_WBC_count	378558	71	2965	0	0.6	0.00	0.96	0.93	0.01	6.43	3037	0.99
2q13_dup	heart_valve	373146	70	8377	1	1.6	0.64	0.97	0.97	0.11	3.56	3069	0.99
2q13_dup	diabetes_insulin_dependent	378929	71	2594	0	0.5	0.00	0.99	0.98	0.01	6.80	3105	1.00
2q13_dup	gout	374432	70	7091	1	1.3	0.76	0.99	1.01	0.11	3.78	3106	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q13del(NPHP1)	varicose_veins	371831	2231	11951	99	71.7	1.37	0.0027	1.38	1.12	1.68	63	0.14
2q13del(NPHP1)	ear	369500	2267	14282	63	87.6	0.73	0.0069	0.72	0.56	0.92	92	0.23
2q13del(NPHP1)	osteoporosis	372095	2236	11687	94	70.2	1.33	0.0073	1.35	1.09	1.66	100	0.23
2q13del(NPHP1)	nasal	380560	2300	3222	30	19.5	1.54	0.022	1.57	1.07	2.20	202	0.34
2q13del(NPHP1)	ischaemic_heart_disease_not_MI	367342	2213	16440	117	99.0	1.17	0.075	1.19	0.98	1.43	427	0.55
2q13del(NPHP1)	obesity	373870	2255	9912	75	59.8	1.25	0.075	1.24	0.98	1.56	428	0.55
2q13del(NPHP1)	arrhythmia	359456	2162	24326	168	146.3	1.14	0.079	1.16	0.98	1.35	446	0.55
2q13del(NPHP1)	endocrine_other	381401	2309	2381	21	14.4	1.46	0.087	1.49	0.94	2.22	478	0.57
2q13del(NPHP1)	MI_derived2017	369823	2231	13959	99	84.2	1.17	0.097	1.20	0.97	1.46	515	0.59
2q13del(NPHP1)	renal_failure	375803	2271	7979	59	48.2	1.22	0.11	1.24	0.95	1.60	566	0.63
2q13del(NPHP1)	allergy	324789	1997	58993	333	362.7	0.93	0.12	0.91	0.81	1.02	612	0.63
2q13del(NPHP1)	cataract	361782	2180	22000	150	132.6	1.12	0.15	1.13	0.95	1.34	702	0.69
2q13del(NPHP1)	congenital	378717	2292	5065	38	30.7	1.24	0.18	1.25	0.90	1.70	776	0.72
2q13del(NPHP1)	anaemia	364539	2227	19243	103	117.6	0.88	0.19	0.88	0.72	1.06	800	0.73
2q13del(NPHP1)	osteoarthritis	314631	1884	69151	446	414.1	1.06	0.23	1.07	0.96	1.19	929	0.77
2q13del(NPHP1)	low_WBC_count	380804	2317	2978	13	18.1	0.72	0.26	0.74	0.41	1.22	1014	0.79
2q13del(NPHP1)	gout	376657	2295	7125	35	43.4	0.81	0.26	0.83	0.58	1.14	1031	0.79
2q13del(NPHP1)	any_Ca	333800	2044	49982	286	306.1	0.94	0.27	0.93	0.82	1.06	1066	0.80
2q13del(NPHP1)	diverticular_disease_intestine	356495	2176	27287	154	166.6	0.93	0.30	0.92	0.77	1.08	1168	0.81
2q13del(NPHP1)	ocular	361979	2187	21803	143	131.7	1.08	0.34	1.09	0.91	1.28	1299	0.83
2q13del(NPHP1)	gastric_reflux	349290	2133	34492	197	210.6	0.94	0.36	0.93	0.80	1.08	1339	0.84
2q13del(NPHP1)	respiratory	362916	2214	20866	116	127.3	0.92	0.36	0.92	0.76	1.10	1352	0.84
2q13del(NPHP1)	venous_thromboembolic_disease	369716	2236	14066	94	85.1	1.10	0.39	1.10	0.89	1.34	1436	0.85
2q13del(NPHP1)	hypertension	262396	1575	121386	755	728.6	1.02	0.40	1.04	0.95	1.14	1483	0.85
2q13del(NPHP1)	irritable_bowel_syndrome	370692	2243	13090	87	79.2	1.10	0.42	1.09	0.88	1.35	1503	0.87
2q13del(NPHP1)	uterine_problem	341219	2055	42563	275	256.3	1.06	0.42	1.06	0.92	1.21	1531	0.87
2q13del(NPHP1)	stroke_derived2017	374905	2271	8877	59	53.8	1.10	0.43	1.11	0.85	1.43	1559	0.87
2q13del(NPHP1)	prostate_hyperplasia	369241	2236	14541	94	88.1	1.07	0.46	1.09	0.87	1.34	1635	0.88
2q13del(NPHP1)	connective_tissue_disorder	374368	2268	9414	62	57.0	1.09	0.51	1.09	0.84	1.39	1792	0.89
2q13del(NPHP1)	hernia	335612	2048	48170	282	293.9	0.96	0.52	0.96	0.84	1.09	1808	0.90
2q13del(NPHP1)	renal	376445	2282	7337	48	44.5	1.08	0.54	1.09	0.81	1.44	1871	0.91
2q13del(NPHP1)	atherosclerotic_vascular_disease	377995	2292	5787	38	35.1	1.08	0.56	1.10	0.79	1.50	1899	0.92
2q13del(NPHP1)	biliary	362886	2208	20896	122	127.1	0.96	0.62	0.96	0.79	1.14	2075	0.94
2q13del(NPHP1)	coagulation_defects	381224	2313	2558	17	15.5	1.10	0.63	1.13	0.68	1.75	2085	0.94
2q13del(NPHP1)	heart_valve	375351	2275	8431	55	51.1	1.08	0.63	1.07	0.81	1.38	2109	0.94
2q13del(NPHP1)	paralytic_syndromes	381812	2320	1970	10	12.0	0.84	0.67	0.88	0.45	1.53	2193	0.95
2q13del(NPHP1)	died_to_2016	372750	2267	11032	63	67.1	0.94	0.69	0.95	0.73	1.21	2263	0.95
2q13del(NPHP1)	COPD	369471	2247	14311	83	87.0	0.96	0.72	0.96	0.76	1.19	2351	0.96
2q13del(NPHP1)	psoriasis	377724	2292	6058	38	36.8	1.03	0.73	1.06	0.75	1.43	2393	0.96
2q13del(NPHP1)	diabetes_other	362925	2207	20857	123	126.8	0.97	0.74	0.97	0.80	1.16	2397	0.96
2q13del(NPHP1)	gastrointestinal_ulcer	373370	2270	10412	60	63.3	0.95	0.79	0.97	0.74	1.24	2553	0.97
2q13del(NPHP1)	glaucoma	377216	2292	6566	38	39.9	0.95	0.80	0.96	0.68	1.30	2570	0.97
2q13del(NPHP1)	ovarian_cysts	376350	2286	7432	44	45.1	0.98	0.81	0.96	0.70	1.28	2606	0.97
2q13del(NPHP1)	high_cholesterol	316792	1920	66990	410	406.0	1.01	0.84	1.01	0.90	1.13	2688	0.97
2q13del(NPHP1)	cerebrovascular_disease_not_stroke	379425	2303	4357	27	26.4	1.02	0.85	1.04	0.69	1.48	2728	0.97
2q13del(NPHP1)	heart_other	371888	2257	11894	73	72.2	1.01	0.85	1.02	0.80	1.28	2740	0.97
2q13del(NPHP1)	inflammatory_bowel_disease	365077	2215	18705	115	113.5	1.01	0.85	1.02	0.84	1.22	2742	0.97
2q13del(NPHP1)	diabetes_insulin_dependent	381172	2314	2610	16	15.8	1.01	0.86	1.04	0.62	1.64	2771	0.98
2q13del(NPHP1)	neuropathies	367783	2234	15999	96	97.2	0.99	0.88	0.98	0.80	1.20	2803	0.98
2q13del(NPHP1)	ureter_bladder	334957	2032	48825	298	296.2	1.01	0.88	1.01	0.89	1.14	2808	0.98
2q13del(NPHP1)	migraine	370021	2247	13761	83	83.6	0.99	0.88	0.98	0.78	1.22	2816	0.98
2q13del(NPHP1)	asthma	334155	2027	49627	303	301.0	1.01	0.88	1.01	0.89	1.14	2823	0.98
2q13del(NPHP1)	aneurism	381804	2318	1978	12	12.0	1.00	0.90	1.04	0.56	1.73	2880	0.98
2q13del(NPHP1)	thyroid	356474	2162	27308	168	165.6	1.01	0.91	1.01	0.86	1.18	2908	0.98
2q13del(NPHP1)	heartfailure	378677	2300	5105	30	31.0	0.97	0.94	0.99	0.67	1.39	2986	0.99
2q13del(NPHP1)	hepatic	374976	2277	8806	53	53.5	0.99	0.96	1.01	0.76	1.31	3027	0.99
2q13del(NPHP1)	digestive	301439	1831	82343	499	500.2	1.00	0.97	1.00	0.90	1.10	3073	0.99
2q13del(NPHP1)	sciatica	340407	2066	43375	264	263.3	1.00	0.97	1.00	0.88	1.14	3076	0.99

CNV	Phenotype	N_con	N_CNV_ con	N_case	N_CNV_ case	Expected_ N_CNVS	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q13dup(NPHP1)	hypertension	262132	1311	121188	557	606.1	0.94	0.055	0.90	0.81	1.00	352	0.49
2q13dup(NPHP1)	respiratory	362450	1748	20870	120	100.7	1.18	0.063	1.20	0.99	1.44	371	0.53
2q13dup(NPHP1)	thyroid	356029	1717	27291	151	131.6	1.14	0.067	1.18	0.99	1.39	384	0.55
2q13dup(NPHP1)	psoriasis	377280	1848	6040	20	29.6	0.68	0.077	0.69	0.43	1.04	436	0.55
2q13dup(NPHP1)	hernia	335218	1654	48102	214	237.3	0.91	0.094	0.89	0.76	1.02	502	0.59
2q13dup(NPHP1)	hepatic	374513	1814	8807	54	42.7	1.26	0.12	1.25	0.94	1.63	601	0.63
2q13dup(NPHP1)	osteoarthritis	314258	1511	69062	357	332.1	1.06	0.16	1.09	0.97	1.23	725	0.70
2q13dup(NPHP1)	ischaemic_heart_disease_not_MI	366928	1799	16392	69	80.4	0.86	0.18	0.85	0.66	1.07	768	0.72
2q13dup(NPHP1)	ovarian_cysts	375889	1825	7431	43	36.1	1.19	0.20	1.23	0.89	1.64	857	0.74
2q13dup(NPHP1)	any_Ca	333400	1644	49920	224	246.2	0.92	0.21	0.91	0.79	1.05	875	0.75
2q13dup(NPHP1)	ear	369021	1788	14299	80	69.3	1.15	0.21	1.16	0.92	1.44	883	0.75
2q13dup(NPHP1)	MI_derived2017	369401	1809	13919	59	68.2	0.87	0.23	0.85	0.65	1.10	933	0.77
2q13dup(NPHP1)	obesity	373443	1828	9877	40	48.3	0.83	0.23	0.83	0.60	1.12	935	0.78
2q13dup(NPHP1)	allergy	324353	1561	58967	307	283.8	1.07	0.27	1.07	0.95	1.21	1051	0.80
2q13dup(NPHP1)	coagulation_defects	380762	1851	2558	17	12.4	1.37	0.28	1.32	0.78	2.07	1102	0.80
2q13dup(NPHP1)	low_WBC_count	380344	1857	2976	11	14.5	0.76	0.28	0.73	0.37	1.27	1104	0.80
2q13dup(NPHP1)	osteoporosis	371664	1805	11656	63	56.6	1.11	0.31	1.14	0.88	1.46	1200	0.81
2q13dup(NPHP1)	renal_failure	375355	1823	7965	45	38.7	1.16	0.32	1.17	0.85	1.55	1235	0.82
2q13dup(NPHP1)	uterine_problem	340841	1677	42479	191	209.0	0.92	0.36	0.93	0.79	1.09	1348	0.85
2q13dup(NPHP1)	cerebrovascular_disease_not_stroke	378973	1851	4347	17	21.2	0.80	0.37	0.81	0.49	1.26	1361	0.85
2q13dup(NPHP1)	nasal	380116	1856	3204	12	15.6	0.77	0.38	0.79	0.43	1.31	1409	0.85
2q13dup(NPHP1)	stroke_derived2017	374465	1831	8855	37	43.3	0.86	0.39	0.87	0.62	1.18	1423	0.85
2q13dup(NPHP1)	venous_thromboembolic_disease	369273	1793	14047	75	68.2	1.10	0.39	1.11	0.87	1.39	1438	0.85
2q13dup(NPHP1)	neuropathies	367346	1797	15974	71	78.1	0.91	0.45	0.91	0.71	1.15	1611	0.87
2q13dup(NPHP1)	gout	376190	1828	7130	40	34.6	1.15	0.45	1.13	0.81	1.53	1614	0.87
2q13dup(NPHP1)	renal	376000	1837	7320	31	35.8	0.87	0.46	0.88	0.60	1.23	1641	0.88
2q13dup(NPHP1)	inflammatory_bowel_disease	364632	1770	18688	98	90.7	1.08	0.47	1.08	0.87	1.32	1682	0.88
2q13dup(NPHP1)	died_to_2016	372303	1820	11017	48	53.9	0.89	0.49	0.90	0.67	1.19	1722	0.89
2q13dup(NPHP1)	arrhythmia	359036	1742	24284	126	117.8	1.07	0.51	1.06	0.88	1.27	1778	0.89
2q13dup(NPHP1)	heart_valve	374907	1831	8413	37	41.1	0.90	0.52	0.90	0.64	1.23	1828	0.90
2q13dup(NPHP1)	atherosclerotic_vascular_disease	377540	1837	5780	31	28.1	1.10	0.55	1.12	0.77	1.56	1880	0.91
2q13dup(NPHP1)	heart_other	371445	1814	11875	54	58.0	0.93	0.56	0.92	0.70	1.20	1926	0.92
2q13dup(NPHP1)	aneurism	381343	1857	1977	11	9.6	1.14	0.59	1.18	0.62	2.01	1973	0.93
2q13dup(NPHP1)	gastric_reflux	348852	1695	34468	173	167.5	1.03	0.60	1.04	0.89	1.22	2017	0.94
2q13dup(NPHP1)	connective_tissue_disorder	373920	1820	9400	48	45.8	1.05	0.61	1.08	0.80	1.42	2034	0.94
2q13dup(NPHP1)	ureter_bladder	334563	1638	48757	230	238.7	0.97	0.62	0.97	0.84	1.11	2063	0.94
2q13dup(NPHP1)	heartfailure	378218	1841	5102	27	24.8	1.09	0.64	1.10	0.73	1.57	2122	0.94
2q13dup(NPHP1)	COPD	369020	1796	14300	72	69.6	1.03	0.66	1.06	0.82	1.33	2175	0.95
2q13dup(NPHP1)	irritable_bowel_syndrome	370251	1802	13069	66	63.6	1.04	0.67	1.06	0.82	1.34	2199	0.95
2q13dup(NPHP1)	asthma	333748	1620	49572	248	240.6	1.03	0.68	1.03	0.90	1.17	2227	0.95
2q13dup(NPHP1)	digestive	301070	1462	82250	406	399.4	1.01	0.70	1.02	0.91	1.14	2288	0.96
2q13dup(NPHP1)	varicose_veins	371414	1814	11906	54	58.1	0.93	0.70	0.95	0.72	1.23	2302	0.96
2q13dup(NPHP1)	biliary	362450	1772	20870	96	102.0	0.94	0.71	0.96	0.78	1.18	2314	0.96
2q13dup(NPHP1)	diverticular_disease_intestine	356051	1732	27269	136	132.6	1.02	0.72	1.03	0.86	1.23	2352	0.96
2q13dup(NPHP1)	sciatica	339993	1652	43327	216	210.5	1.02	0.76	1.02	0.88	1.18	2454	0.96
2q13dup(NPHP1)	endocrine_other	380948	1856	2372	12	11.6	1.04	0.76	1.09	0.59	1.82	2474	0.96
2q13dup(NPHP1)	anemia	364089	1777	19231	91	93.9	0.97	0.76	0.97	0.78	1.19	2478	0.96
2q13dup(NPHP1)	cataract	361361	1759	21959	109	106.9	1.02	0.81	1.02	0.84	1.24	2611	0.97
2q13dup(NPHP1)	diabetes_other	362481	1763	20839	105	101.4	1.03	0.82	1.02	0.83	1.24	2628	0.97
2q13dup(NPHP1)	ocular	361552	1760	21768	108	106.0	1.02	0.82	1.02	0.84	1.24	2654	0.97
2q13dup(NPHP1)	prostate_hyperplasia	368797	1792	14523	76	70.6	1.07	0.83	1.03	0.80	1.30	2669	0.97
2q13dup(NPHP1)	glaucoma	376761	1837	6559	31	32.0	0.97	0.85	0.97	0.66	1.35	2725	0.97
2q13dup(NPHP1)	high_cholesterol	316411	1539	66909	329	325.4	1.01	0.86	0.99	0.87	1.12	2769	0.97
2q13dup(NPHP1)	diabetes_insulin_dependent	380713	1855	2607	13	12.7	1.02	0.86	1.05	0.58	1.72	2775	0.98
2q13dup(NPHP1)	migraine	369577	1803	13743	65	67.0	0.97	0.88	0.98	0.76	1.25	2810	0.98
2q13dup(NPHP1)	congenital	378268	1843	5052	25	24.6	1.02	0.88	1.03	0.68	1.49	2812	0.98
2q13dup(NPHP1)	gastrointestinal_ulcer	372917	1817	10403	51	50.7	1.01	0.89	1.02	0.76	1.33	2840	0.98
2q13dup(NPHP1)	paralytic_syndromes	381351	1859	1969	9	9.6	0.94	0.98	0.99	0.48	1.77	3077	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVS	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q21.1 del	aneurism	379524	38	1968	2	0.2	9.70	0.0069	11.20	2.26	34.54	94	0.23
2q21.1 del	gastrointestinal_ulcer	371136	36	10356	4	1.0	3.68	0.015	4.35	1.39	10.59	157	0.30
2q21.1 del	sciatica	338372	31	43120	9	4.0	1.99	0.040	2.28	1.04	4.55	293	0.43
2q21.1 del	COPD	367260	36	14232	4	1.4	2.68	0.072	2.86	0.90	7.15	415	0.54
2q21.1 del	paralytic syndromes	379531	39	1961	1	0.2	4.87	0.078	6.68	0.75	25.12	444	0.55
2q21.1 del	atherosclerotic_vascular_disease	375741	38	5751	2	0.6	3.32	0.086	3.89	0.79	11.76	472	0.57
2q21.1 del	hernia	333595	31	47897	9	4.5	1.79	0.087	1.99	0.90	4.05	479	0.57
2q21.1 del	endocrine_other	379131	39	2361	1	0.2	4.04	0.092	6.03	0.68	22.57	495	0.58
2q21.1 del	renal	374201	38	7291	2	0.7	2.62	0.13	3.21	0.66	9.56	631	0.64
2q21.1 del	any_Ca	331794	38	49698	2	5.7	0.38	0.14	0.42	0.09	1.27	665	0.66
2q21.1 del	hypertension	260844	23	120648	17	10.6	1.34	0.20	1.55	0.79	2.96	827	0.74
2q21.1 del	hepatic	372737	38	8755	2	0.9	2.18	0.20	2.60	0.53	7.75	848	0.74
2q21.1 del	respiratory	360738	36	20754	4	2.1	1.84	0.21	2.01	0.64	4.87	863	0.74
2q21.1 del	cerebrovascular_disease_not_stroke	377161	39	4331	1	0.4	2.20	0.24	3.17	0.36	12.05	969	0.78
2q21.1 del	prostate_hyperplasia	367042	37	14450	3	1.5	1.98	0.24	2.13	0.55	6.24	980	0.78
2q21.1 del	osteoarthritis	312777	30	68715	10	6.6	1.39	0.25	1.56	0.72	3.12	1000	0.78
2q21.1 del	MI_derived2017	367632	40	13860	0	1.5	0.00	0.26	0.28	0.00	2.00	1042	0.80
2q21.1 del	uterine problem	339198	34	42294	6	4.2	1.35	0.27	1.72	0.63	4.18	1050	0.80
2q21.1 del	ischaemic_heart_disease_not_MI	365166	37	16326	3	1.7	1.75	0.29	1.92	0.52	5.18	1129	0.81
2q21.1 del	congenital	376464	39	5028	1	0.5	1.90	0.29	2.76	0.31	10.28	1136	0.81
2q21.1 del	high_cholesterol	314907	35	66585	5	7.4	0.72	0.31	0.63	0.22	1.48	1184	0.81
2q21.1 del	arrhythmia	357330	36	24162	4	2.4	1.58	0.32	1.72	0.55	4.21	1222	0.82
2q21.1 del	thyroid	354348	36	27144	4	2.8	1.41	0.33	1.71	0.54	4.26	1243	0.82
2q21.1 del	inflammatory_bowel_disease	362899	37	18593	3	1.9	1.54	0.35	1.76	0.48	4.61	1313	0.83
2q21.1 del	irritable_bowel_syndrome	368489	40	13003	0	1.4	0.00	0.37	0.35	0.00	2.45	1378	0.85
2q21.1 del	asthma	332161	33	49331	7	4.9	1.35	0.38	1.45	0.61	3.04	1390	0.85
2q21.1 del	anaemia	362349	37	19143	3	2.0	1.49	0.38	1.70	0.46	4.47	1398	0.85
2q21.1 del	psoriasis	375471	39	6021	1	0.6	1.58	0.38	2.28	0.26	8.52	1403	0.85
2q21.1 del	migraine	367812	38	13680	2	1.4	1.39	0.41	1.81	0.37	5.43	1488	0.86
2q21.1 del	osteoporosis	369899	40	11593	0	1.3	0.00	0.45	0.39	0.00	2.88	1604	0.87
2q21.1 del	diabetes_other	360757	39	20735	1	2.2	0.46	0.46	0.57	0.06	2.16	1648	0.88
2q21.1 del	cataract	359639	37	21853	3	2.2	1.31	0.52	1.48	0.39	4.06	1819	0.90
2q21.1 del	ocular	359831	39	21661	1	2.3	0.44	0.52	0.61	0.07	2.29	1829	0.90
2q21.1 del	digestive	299638	30	81854	10	8.2	1.17	0.59	1.22	0.57	2.40	1988	0.93
2q21.1 del	renal_failure	373571	39	7921	1	0.8	1.20	0.60	1.60	0.18	6.07	2015	0.94
2q21.1 del	heart_valve	373116	40	8376	0	0.9	0.00	0.61	0.52	0.00	3.66	2030	0.94
2q21.1 del	connective_tissue_disorder	372139	39	9353	1	1.0	1.02	0.66	1.48	0.17	5.61	2167	0.95
2q21.1 del	gout	374402	40	7090	0	0.8	0.00	0.66	0.57	0.00	4.09	2184	0.95
2q21.1 del	stroke_derived2017	372673	39	8819	1	0.9	1.08	0.67	1.45	0.16	5.52	2218	0.95
2q21.1 del	ovarian_cysts	374104	40	7388	0	0.8	0.00	0.72	0.62	0.00	4.50	2329	0.96
2q21.1 del	allergy	322827	35	58665	5	6.4	0.81	0.72	0.85	0.31	1.93	2343	0.96
2q21.1 del	coagulation_defects	378951	40	2541	0	0.3	0.00	0.72	1.74	0.01	12.20	2348	0.96
2q21.1 del	glaucoma	374964	40	6528	0	0.7	0.00	0.75	0.65	0.01	4.65	2431	0.97
2q21.1 del	diverticular_disease_intestine	354356	37	27136	3	2.8	1.05	0.75	1.20	0.32	3.19	2443	0.97
2q21.1 del	low_WBC_count	378527	40	2965	0	0.3	0.00	0.76	1.60	0.01	11.19	2463	0.97
2q21.1 del	diabetes_insulin_dependent	378898	40	2594	0	0.3	0.00	0.76	1.59	0.01	11.20	2477	0.96
2q21.1 del	obesity	371654	39	9838	1	1.0	0.97	0.76	1.30	0.15	4.87	2480	0.97
2q21.1 del	neuropathies	365588	39	15904	1	1.7	0.60	0.83	0.84	0.09	3.14	2672	0.97
2q21.1 del	nasal	378300	40	3192	0	0.3	0.00	0.83	1.37	0.01	9.67	2679	0.97
2q21.1 del	varicose_veins	369639	39	11853	1	1.3	0.80	0.84	1.19	0.13	4.44	2712	0.97
2q21.1 del	died_to_2016	370522	39	10970	1	1.2	0.87	0.85	1.17	0.13	4.40	2749	0.97
2q21.1 del	biliary	360716	38	20776	2	2.2	0.92	0.86	1.12	0.23	3.36	2773	0.98
2q21.1 del	heartfailure	376417	40	5075	0	0.5	0.00	0.87	0.80	0.01	5.76	2796	0.98
2q21.1 del	gastric_reflux	347194	37	34298	3	3.7	0.83	0.87	0.92	0.25	2.40	2797	0.98
2q21.1 del	heart_other	369670	39	11822	1	1.2	0.81	0.93	1.08	0.12	4.09	2952	0.98
2q21.1 del	ear	367272	39	14220	1	1.5	0.67	0.93	0.93	0.10	3.48	2955	0.99
2q21.1 del	venous_thromboembolic_disease	367519	39	13973	1	1.5	0.68	0.95	0.95	0.11	3.57	3022	0.99
2q21.1 del	ureter_bladder	332960	35	48532	5	5.1	0.98	0.95	1.03	0.37	2.34	3026	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q21.1_dup	endocrine_other	379145	53	2362	2	0.3	5.88	0.016	7.85	1.62	23.01	161	0.30
2q21.1_dup	sciatica	338395	54	43112	1	6.9	0.16	0.016	0.22	0.02	0.80	165	0.31
2q21.1_dup	died_to_2016	370533	50	10974	5	1.5	3.16	0.021	3.37	1.23	7.51	193	0.35
2q21.1_dup	asthma	332180	52	49327	3	7.7	0.42	0.095	0.44	0.12	1.13	507	0.59
2q21.1_dup	uterine_problem	339211	47	42296	8	5.9	1.31	0.10	2.07	0.85	4.68	535	0.60
2q21.1_dup	ischaemic_heart_disease_not_MI	365184	55	16323	0	2.5	0.00	0.12	0.19	0.00	1.34	577	0.63
2q21.1_dup	allergy	322835	43	58672	12	7.8	1.42	0.13	1.66	0.85	3.04	649	0.64
2q21.1_dup	gastrointestinal_ulcer	371152	52	10355	3	1.5	2.01	0.18	2.33	0.64	6.03	765	0.72
2q21.1_dup	diverticular_disease_intestine	354368	49	27139	6	3.8	1.53	0.18	1.84	0.73	3.95	777	0.72
2q21.1_dup	arrhythmia	357348	54	24159	1	3.7	0.29	0.18	0.39	0.04	1.44	794	0.73
2q21.1_dup	low_WBC_count	378541	54	2966	1	0.4	2.34	0.19	3.66	0.41	13.47	818	0.74
2q21.1_dup	diabetes_other	360772	54	20735	1	3.1	0.33	0.25	0.43	0.05	1.60	984	0.78
2q21.1_dup	heart_other	369683	52	11824	3	1.7	1.76	0.27	1.95	0.53	5.09	1075	0.80
2q21.1_dup	inflammatory_bowel_disease	362913	51	18594	4	2.6	1.49	0.31	1.72	0.56	4.06	1178	0.81
2q21.1_dup	renal_failure	373585	53	7922	2	1.1	1.75	0.31	2.10	0.43	6.25	1182	0.81
2q21.1_dup	gout	374415	53	7092	2	1.0	1.96	0.31	2.08	0.42	6.26	1212	0.81
2q21.1_dup	biliary	360729	51	20778	4	2.9	1.34	0.34	1.66	0.54	3.93	1290	0.83
2q21.1_dup	osteoporosis	369912	53	11595	2	1.7	1.20	0.36	1.95	0.40	5.93	1333	0.84
2q21.1_dup	ear	367285	52	14222	3	2.0	1.46	0.36	1.72	0.47	4.42	1347	0.85
2q21.1_dup	osteoarthritis	312795	48	68712	7	10.5	0.71	0.43	0.74	0.31	1.52	1544	0.87
2q21.1_dup	prostate_hyperplasia	367059	54	14448	1	2.1	0.48	0.44	0.55	0.06	2.15	1589	0.87
2q21.1_dup	heart_valve	373131	55	8376	0	1.2	0.00	0.46	0.41	0.00	2.82	1629	0.88
2q21.1_dup	any_Ca	331806	50	49701	5	7.5	0.70	0.53	0.76	0.28	1.69	1834	0.90
2q21.1_dup	MI_derived2017	367646	54	13861	1	2.0	0.50	0.55	0.63	0.07	2.39	1896	0.92
2q21.1_dup	ovarian_cysts	374118	54	7389	1	1.1	0.94	0.55	1.71	0.19	6.63	1897	0.92
2q21.1_dup	psoriasis	375486	54	6021	1	0.9	1.15	0.57	1.65	0.19	6.09	1943	0.92
2q21.1_dup	glaucoma	374978	54	6529	1	0.9	1.06	0.59	1.62	0.18	6.06	1980	0.93
2q21.1_dup	hypertension	260860	39	120647	16	18.0	0.92	0.64	0.87	0.47	1.55	2123	0.95
2q21.1_dup	COPD	367278	54	14229	1	2.1	0.49	0.64	0.69	0.08	2.60	2133	0.94
2q21.1_dup	anaemia	362364	52	19143	3	2.7	1.09	0.64	1.30	0.36	3.36	2136	0.94
2q21.1_dup	atherosclerotic_vascular_disease	375758	55	5749	0	0.8	0.00	0.66	0.57	0.00	3.98	2185	0.95
2q21.1_dup	venous_thromboembolic_disease	367534	54	13973	1	2.1	0.50	0.69	0.73	0.08	2.69	2264	0.95
2q21.1_dup	heartfailure	376432	55	5075	0	0.7	0.00	0.70	0.60	0.00	4.25	2281	0.96
2q21.1_dup	ocular	359845	53	21662	2	3.2	0.64	0.70	0.79	0.16	2.31	2298	0.96
2q21.1_dup	renal	374217	54	7290	1	1.1	0.95	0.71	1.38	0.16	5.09	2310	0.96
2q21.1_dup	hernia	333613	49	47894	6	7.0	0.87	0.72	0.86	0.34	1.84	2338	0.96
2q21.1_dup	congenital	376480	55	5027	0	0.7	0.00	0.75	0.66	0.01	4.54	2426	0.97
2q21.1_dup	aneurism	379541	55	1966	0	0.3	0.00	0.76	1.59	0.01	11.21	2479	0.96
2q21.1_dup	paralytic_syndromes	379547	55	1960	0	0.3	0.00	0.77	1.57	0.01	10.95	2489	0.97
2q21.1_dup	ureter_bladder	332974	49	48533	6	7.1	0.86	0.77	0.89	0.35	1.88	2494	0.97
2q21.1_dup	cataract	359655	53	21852	2	3.2	0.63	0.77	0.83	0.17	2.49	2498	0.97
2q21.1_dup	gastric_reflux	347207	50	34300	5	4.9	1.01	0.80	1.12	0.41	2.48	2601	0.97
2q21.1_dup	migraine	367828	54	13679	1	2.0	0.51	0.82	0.83	0.09	3.09	2643	0.97
2q21.1_dup	connective_tissue_disorder	372154	54	9353	1	1.4	0.74	0.84	1.19	0.13	4.42	2684	0.97
2q21.1_dup	irritable_bowel_syndrome	368503	54	13004	1	1.9	0.53	0.84	0.85	0.10	3.14	2700	0.97
2q21.1_dup	coagulation_defects	378966	55	2541	0	0.4	0.00	0.86	1.31	0.01	9.10	2754	0.97
2q21.1_dup	cerebrovascular_disease_not_stroke	377177	55	4330	0	0.6	0.00	0.86	0.79	0.01	5.55	2776	0.98
2q21.1_dup	high_cholesterol	314917	45	66590	10	9.5	1.04	0.87	1.06	0.50	2.06	2798	0.98
2q21.1_dup	neuropathies	365602	53	15905	2	2.3	0.87	0.88	1.11	0.23	3.25	2804	0.98
2q21.1_dup	respiratory	360754	52	20753	3	3.0	1.00	0.88	1.09	0.30	2.81	2814	0.98
2q21.1_dup	digestive	299651	43	81856	12	11.7	1.02	0.89	1.05	0.53	1.92	2831	0.98
2q21.1_dup	hepatic	372753	54	8754	1	1.3	0.79	0.90	1.12	0.13	4.11	2867	0.98
2q21.1_dup	varicose_veins	369654	54	11853	1	1.7	0.59	0.91	0.91	0.10	3.35	2896	0.98
2q21.1_dup	diabetes_insulin_dependent	378913	55	2594	0	0.4	0.00	0.91	1.18	0.01	8.22	2902	0.98
2q21.1_dup	stroke_derived2017	372688	54	8819	1	1.3	0.79	0.91	1.09	0.12	4.08	2912	0.98
2q21.1_dup	thyroid	354364	52	27143	3	4.0	0.77	0.95	1.03	0.28	2.72	3019	0.99
2q21.1_dup	nasal	378315	55	3192	0	0.5	0.00	0.97	1.05	0.01	7.33	3070	0.99
2q21.1_dup	obesity	371669	54	9838	1	1.4	0.71	1.00	1.00	0.11	3.68	3123	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
3q29_del	glaucoma	374931	7	6530	2	0.1	12.99	0.0016	23.02	4.14	90.99	137	0.10
3q29_del	irritable_bowel_syndrome	368456	7	13005	2	0.2	6.52	0.012	10.06	1.83	39.47	137	0.28
3q29_del	cataract	359609	7	21852	2	0.4	3.88	0.029	7.29	1.28	29.95	239	0.37
3q29_del	heartfailure	376385	8	5076	1	0.1	8.35	0.034	12.56	1.28	61.19	259	0.41
3q29_del	high_cholesterol	314877	5	66584	4	1.1	2.55	0.036	4.41	1.11	16.54	271	0.41
3q29_del	ocular	359799	7	21662	2	0.4	3.91	0.042	5.84	1.08	22.24	298	0.44
3q29_del	diabetes_other	360725	7	20736	2	0.4	4.09	0.049	5.57	1.01	21.91	326	0.47
3q29_del	arrhythmia	357301	7	24160	2	0.5	3.51	0.056	5.24	0.95	20.48	354	0.50
3q29_del	renal	374711	8	7290	1	0.2	5.81	0.058	8.56	0.91	38.25	359	0.51
3q29_del	renal_failure	373540	8	7921	1	0.2	5.35	0.069	7.87	0.82	36.81	394	0.54
3q29_del	asthma	332134	6	49327	3	0.9	2.58	0.073	3.66	0.87	12.94	421	0.55
3q29_del	stroke_derived2017	372642	8	8819	1	0.2	4.81	0.083	6.91	0.72	32.18	465	0.56
3q29_del	allergy	322798	6	58663	3	1.1	2.17	0.10	3.28	0.77	11.68	528	0.60
3q29_del	heart_other	369639	8	11822	1	0.3	3.59	0.12	5.41	0.56	25.38	602	0.63
3q29_del	venous_thromboembolic_disease	367488	8	13973	1	0.3	3.03	0.15	4.57	0.48	20.61	697	0.68
3q29_del	ear	367241	8	14220	1	0.3	2.98	0.16	4.50	0.48	20.14	704	0.69
3q29_del	MI_derived2017	367600	8	13861	1	0.3	3.06	0.17	4.42	0.45	22.01	746	0.71
3q29_del	ischaemic_heart_disease_not_MI	365137	8	16324	1	0.4	2.60	0.19	4.08	0.42	19.47	797	0.73
3q29_del	osteoarthritis	312756	9	68705	0	2.0	0.00	0.23	0.24	0.00	1.96	921	0.77
3q29_del	aneurism	379495	9	1966	0	0.0	0.00	0.24	10.44	0.08	90.55	948	0.78
3q29_del	sciatica	338348	7	43113	2	0.9	1.97	0.24	2.54	0.47	9.55	977	0.78
3q29_del	anaemia	362320	8	19141	1	0.4	2.21	0.25	3.28	0.35	14.76	983	0.78
3q29_del	inflammatory_bowel_disease	362870	8	18591	1	0.4	2.28	0.25	3.26	0.35	14.57	986	0.78
3q29_del	biliary	360686	8	20775	1	0.5	2.04	0.25	3.24	0.34	14.83	1006	0.78
3q29_del	endocrine_other	379101	9	2360	0	0.1	0.00	0.27	8.26	0.06	66.05	1060	0.80
3q29_del	paralytic_syndromes	379501	9	1960	0	0.0	0.00	0.27	8.22	0.06	65.71	1063	0.80
3q29_del	coagulation_defects	378920	9	2541	0	0.1	0.00	0.29	7.39	0.06	58.44	1128	0.81
3q29_del	nasal	378269	9	3192	0	0.1	0.00	0.30	7.13	0.06	56.79	1151	0.81
3q29_del	low_WBC_count	378496	9	2965	0	0.1	0.00	0.30	7.04	0.05	55.64	1160	0.81
3q29_del	diabetes_insulin_dependent	378867	9	2594	0	0.1	0.00	0.32	6.49	0.05	51.98	1215	0.82
3q29_del	thyroid	354320	8	27141	1	0.6	1.56	0.34	2.67	0.27	13.06	1273	0.83
3q29_del	diverticular_disease_intestine	354327	8	27134	1	0.6	1.56	0.34	2.59	0.27	11.92	1295	0.83
3q29_del	cerebrovascular_disease_not_stroke	377131	9	4330	0	0.1	0.00	0.38	4.86	0.04	40.35	1415	0.85
3q29_del	hernia	333573	9	47888	0	1.3	0.00	0.38	0.34	0.00	2.75	1417	0.85
3q29_del	hypertension	260826	5	120635	4	2.3	1.41	0.39	1.78	0.46	6.54	1435	0.85
3q29_del	congenital	376434	9	5027	0	0.1	0.00	0.46	3.63	0.03	28.64	1632	0.88
3q29_del	atherosclerotic_vascular_disease	375712	9	5749	0	0.1	0.00	0.49	3.23	0.02	26.26	1744	0.89
3q29_del	psoriasis	375441	9	6020	0	0.1	0.00	0.51	3.06	0.02	24.17	1787	0.89
3q29_del	ovarian_cysts	374073	9	7388	0	0.2	0.00	0.55	2.80	0.02	26.31	1874	0.91
3q29_del	gastric_reflux	347165	8	34296	1	0.8	1.24	0.55	1.77	0.19	7.92	1881	0.91
3q29_del	digestive	299616	8	81845	1	2.2	0.52	0.56	0.61	0.06	2.75	1922	0.92
3q29_del	heart_valve	373085	9	8376	0	0.2	0.00	0.59	2.41	0.02	19.44	1995	0.93
3q29_del	gout	374371	9	7090	0	0.2	0.00	0.61	2.35	0.02	20.56	2027	0.94
3q29_del	connective_tissue_disorder	372109	9	9352	0	0.2	0.00	0.64	2.13	0.02	17.25	2127	0.95
3q29_del	osteoporosis	369868	9	11593	0	0.3	0.00	0.66	2.06	0.02	18.06	2174	0.95
3q29_del	uterine_problem	339172	8	42289	1	1.0	1.00	0.67	1.54	0.15	9.37	2206	0.95
3q29_del	hepatic	372708	9	8753	0	0.2	0.00	0.67	1.97	0.02	15.61	2215	0.95
3q29_del	gastrointestinal_ulcer	371109	9	10352	0	0.3	0.00	0.72	1.75	0.01	14.08	2360	0.96
3q29_del	obesity	371624	9	9837	0	0.2	0.00	0.72	1.75	0.01	13.90	2363	0.96
3q29_del	varicose_veins	369609	9	11852	0	0.3	0.00	0.75	1.64	0.01	13.02	2445	0.96
3q29_del	any_Ca	331764	8	49697	1	1.2	0.85	0.77	1.30	0.14	5.92	2506	0.97
3q29_del	died_to_2016	370492	9	10969	0	0.3	0.00	0.78	1.53	0.01	12.28	2536	0.97
3q29_del	migraine	367783	9	13678	0	0.3	0.00	0.79	1.52	0.01	12.47	2555	0.97
3q29_del	COPD	367233	9	14228	0	0.3	0.00	0.86	1.32	0.01	10.84	2755	0.97
3q29_del	ureter_bladder	332933	8	48528	1	1.2	0.87	0.86	1.17	0.12	5.27	2767	0.98
3q29_del	respiratory	360711	9	20750	0	0.5	0.00	0.89	0.82	0.01	6.52	2826	0.98
3q29_del	neuropathies	365558	9	15903	0	0.4	0.00	0.92	1.17	0.01	9.34	2921	0.98
3q29_del	prostate_hyperplasia	367014	9	14447	0	0.4	0.00	0.94	1.12	0.01	11.66	2989	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
3q29_dup	any_Ca	331757	1	49700	4	0.15	6.14	6.23E-05	37.54	6.51	389.10	17	0.011
3q29_dup	diverticular_disease_intestine	354321	2	27136	3	0.15	8.44	0.00010	41.76	7.38	276.02	19	0.017
3q29_dup	inflammatory_bowel_disease	362864	2	18593	3	0.10	12.31	0.00013	35.45	6.68	217.84	20	0.021
3q29_dup	renal_failure	373535	3	7922	2	0.06	19.27	0.00022	58.45	9.22	324.78	25	0.027
3q29_dup	died_to_2016	370486	3	10971	2	0.09	13.91	0.0013	27.81	4.54	146.05	44	0.093
3q29_dup	neuropathies	365552	3	15905	2	0.13	9.59	0.0019	24.65	3.94	131.53	54	0.11
3q29_dup	low_WBC_count	378491	4	2966	1	0.03	25.73	0.0034	54.22	5.38	297.82	69	0.16
3q29_dup	connective_tissue_disorder	372104	4	9353	1	0.10	8.16	0.014	24.49	2.23	150.02	150	0.30
3q29_dup	COPD	367228	4	14229	1	0.15	5.36	0.020	18.81	1.79	112.34	179	0.35
3q29_dup	obesity	371619	4	9838	1	0.11	7.76	0.021	17.37	1.72	95.51	191	0.35
3q29_dup	hepatic	372703	4	8754	1	0.09	8.72	0.025	15.80	1.56	88.19	215	0.36
3q29_dup	varicose_veins	369604	4	11853	1	0.13	6.44	0.026	15.47	1.53	85.75	221	0.36
3q29_dup	migraine	367778	4	13679	1	0.15	5.58	0.040	11.94	1.16	67.58	289	0.43
3q29_dup	ear	367237	4	14220	1	0.15	5.37	0.044	11.03	1.09	61.03	305	0.45
3q29_dup	anaemia	362316	4	19141	1	0.21	3.99	0.067	8.54	0.83	48.52	386	0.54
3q29_dup	diabetes_other	360722	4	20735	1	0.23	3.68	0.069	8.24	0.81	46.69	400	0.54
3q29_dup	uterine_problem	339168	4	42289	1	0.50	1.80	0.078	1759.05	0.72	18361.09	443	0.55
3q29_dup	aneurism	379491	5	1966	0	0.03	0.00	0.12	33.31	0.25	310.45	575	0.63
3q29_dup	digestive	299611	3	81846	2	0.82	1.86	0.16	3.56	0.58	18.94	710	0.69
3q29_dup	endocrine_other	379097	5	2360	0	0.03	0.00	0.16	20.43	0.15	188.85	711	0.69
3q29_dup	paralytic_syndromes	379497	5	1960	0	0.03	0.00	0.16	18.57	0.14	168.98	735	0.70
3q29_dup	coagulation_defects	378916	5	2541	0	0.03	0.00	0.19	15.10	0.11	135.65	802	0.73
3q29_dup	cerebrovascular_disease_not_stroke	377127	5	4330	0	0.06	0.00	0.19	14.76	0.11	138.15	810	0.74
3q29_dup	diabetes_insulin_dependent	378863	5	2594	0	0.03	0.00	0.20	13.53	0.10	121.36	845	0.74
3q29_dup	heartfailure	376382	5	5075	0	0.07	0.00	0.21	12.97	0.10	118.28	866	0.75
3q29_dup	nasal	378265	5	3192	0	0.04	0.00	0.23	10.64	0.08	97.31	942	0.78
3q29_dup	glaucoma	374929	5	6528	0	0.09	0.00	0.26	8.95	0.07	86.01	1034	0.79
3q29_dup	atherosclerotic_vascular_disease	375708	5	5749	0	0.08	0.00	0.26	8.82	0.07	83.24	1041	0.80
3q29_dup	ovarian_cysts	374069	5	7388	0	0.10	0.00	0.28	8.99	0.06	170.61	1076	0.80
3q29_dup	congenital	376430	5	5027	0	0.07	0.00	0.29	7.35	0.06	65.15	1140	0.81
3q29_dup	hypertension	260824	3	120633	2	1.39	1.26	0.31	2.45	0.39	13.56	1205	0.81
3q29_dup	stroke_derived2017	372639	5	8818	0	0.12	0.00	0.33	6.28	0.05	57.40	1247	0.82
3q29_dup	heart_valve	373081	5	8376	0	0.11	0.00	0.35	5.61	0.04	51.88	1320	0.84
3q29_dup	renal	374168	5	7289	0	0.10	0.00	0.36	5.48	0.04	49.40	1332	0.84
3q29_dup	psoriasis	375437	5	6020	0	0.08	0.00	0.36	5.39	0.04	48.12	1338	0.84
3q29_dup	gout	374367	5	7090	0	0.09	0.00	0.38	5.04	0.04	47.25	1395	0.85
3q29_dup	asthma	332132	4	49325	1	0.59	1.55	0.41	2.32	0.23	12.57	1489	0.86
3q29_dup	gastrointestinal_ulcer	371105	5	10352	0	0.14	0.00	0.41	4.42	0.03	40.57	1492	0.86
3q29_dup	heart_other	369636	5	11821	0	0.16	0.00	0.42	4.26	0.03	38.56	1511	0.87
3q29_dup	MI_derived2017	367597	5	13860	0	0.19	0.00	0.43	4.09	0.03	37.35	1547	0.87
3q29_dup	prostate_hyperplasia	367010	5	14447	0	0.20	0.00	0.44	4.08	0.03	44.96	1569	0.87
3q29_dup	osteoporosis	369864	5	11593	0	0.16	0.00	0.45	4.12	0.03	72.24	1609	0.87
3q29_dup	ischaemic_heart_disease_not_MI	365134	5	16323	0	0.22	0.00	0.46	3.64	0.03	34.02	1649	0.88
3q29_dup	venous_thromboembolic_disease	367485	5	13972	0	0.19	0.00	0.49	3.38	0.03	30.89	1720	0.88
3q29_dup	irritable_bowel_syndrome	368454	5	13003	0	0.18	0.00	0.50	3.29	0.02	31.43	1749	0.89
3q29_dup	cataract	359607	5	21850	0	0.30	0.00	0.54	2.94	0.02	33.18	1855	0.91
3q29_dup	biliary	360683	5	20774	0	0.29	0.00	0.58	2.54	0.02	24.48	1961	0.93
3q29_dup	allergy	322797	5	58660	0	0.91	0.00	0.61	0.51	0.00	4.52	2053	0.94
3q29_dup	ocular	359797	5	21660	0	0.30	0.00	0.65	2.11	0.02	19.42	2153	0.94
3q29_dup	arrhythmia	357299	5	24158	0	0.34	0.00	0.66	2.06	0.02	18.79	2177	0.95
3q29_dup	respiratory	360707	5	20750	0	0.29	0.00	0.67	2.00	0.02	17.96	2212	0.95
3q29_dup	thyroid	354317	5	27140	0	0.38	0.00	0.73	1.79	0.01	22.57	2380	0.96
3q29_dup	high_cholesterol	314877	5	66580	0	1.06	0.00	0.77	0.66	0.00	6.30	2496	0.97
3q29_dup	osteoarthritis	312752	5	68705	0	1.10	0.00	0.83	0.73	0.01	7.23	2675	0.97
3q29_dup	hernia	333569	5	47888	0	0.72	0.00	0.86	0.78	0.01	7.00	2764	0.97
3q29_dup	ureter_bladder	332930	5	48527	0	0.73	0.00	0.89	0.82	0.01	7.37	2836	0.98
3q29_dup	sciatica	338346	5	43111	0	0.64	0.00	0.89	0.82	0.01	7.46	2854	0.98
3q29_dup	gastric_reflux	347162	5	34295	0	0.49	0.00	0.91	1.19	0.01	10.80	2906	0.98



CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
7q11.23_dup_distal	digestive	299631	23	81844	0	6.3	0.00	0.0064	0.08	0.00	0.60	89	0.23
7q11.23_dup_distal	died to 2016	370503	20	10972	3	0.6	4.54	0.010	6.34	1.67	17.81	126	0.26
7q11.23_dup_distal	glaucoma	374945	21	6530	2	0.4	5.08	0.021	7.24	1.43	23.26	192	0.35
7q11.23_dup_distal	stroke_derived2017	372655	21	8820	2	0.5	3.76	0.039	5.65	1.12	18.14	282	0.43
7q11.23_dup_distal	uterine problem	339187	23	42288	0	2.9	0.00	0.060	0.14	0.00	1.06	364	0.52
7q11.23_dup_distal	hernia	333587	23	47888	0	3.3	0.00	0.071	0.15	0.00	1.12	410	0.54
7q11.23_dup_distal	congenital	376447	22	5028	1	0.3	3.30	0.12	5.13	0.57	19.86	595	0.63
7q11.23_dup_distal	arrhythmia	357314	20	24161	3	1.4	2.06	0.12	2.79	0.73	7.92	609	0.63
7q11.23_dup_distal	venous thromboembolic disease	367501	21	13974	2	0.8	2.37	0.14	3.18	0.64	9.97	660	0.65
7q11.23_dup_distal	allergy	322814	22	58661	1	4.0	0.28	0.15	0.35	0.04	1.37	693	0.68
7q11.23_dup_distal	renal failure	373554	22	7921	1	0.5	2.09	0.21	3.50	0.38	13.98	888	0.76
7q11.23_dup_distal	heart valve	373098	22	8377	1	0.5	1.98	0.24	3.23	0.36	12.67	957	0.78
7q11.23_dup_distal	ovarian cysts	374086	22	7389	1	0.4	2.24	0.25	3.20	0.35	13.27	990	0.78
7q11.23_dup_distal	hepatic	372721	22	8754	1	0.5	1.89	0.25	3.07	0.34	11.94	1013	0.79
7q11.23_dup_distal	diabetes_other	360739	21	20736	2	1.2	1.60	0.29	2.23	0.44	7.13	1117	0.80
7q11.23_dup_distal	respiratory	360723	21	20752	2	1.2	1.60	0.29	2.21	0.44	6.96	1118	0.80
7q11.23_dup_distal	diverticular disease intestine	354342	23	27133	0	1.8	0.00	0.30	0.30	0.00	2.19	1164	0.81
7q11.23_dup_distal	osteoporosis	369881	22	11594	1	0.7	1.43	0.37	2.37	0.26	9.82	1379	0.85
7q11.23_dup_distal	high cholesterol	314890	18	66585	5	3.8	1.25	0.38	1.60	0.53	4.10	1391	0.85
7q11.23_dup_distal	heart other	369653	22	11822	1	0.7	1.40	0.40	2.25	0.25	8.99	1462	0.85
7q11.23_dup_distal	paralytic syndromes	379515	23	1960	0	0.1	0.00	0.40	4.51	0.04	32.79	1463	0.85
7q11.23_dup_distal	varicose veins	369622	22	11853	1	0.7	1.40	0.42	2.14	0.24	8.32	1509	0.87
7q11.23_dup_distal	aneurism	379509	23	1966	0	0.1	0.00	0.42	4.09	0.03	31.53	1534	0.87
7q11.23_dup_distal	COPD	367246	22	14229	1	0.9	1.17	0.44	2.06	0.23	8.26	1595	0.87
7q11.23_dup_distal	endocrine other	379115	23	2360	0	0.1	0.00	0.46	3.55	0.03	25.66	1645	0.88
7q11.23_dup_distal	MI_derived2017	367614	22	13861	1	0.8	1.20	0.48	1.95	0.21	8.16	1712	0.88
7q11.23_dup_distal	diabetes insulin dependent	378881	23	2594	0	0.2	0.00	0.48	3.29	0.03	23.83	1719	0.88
7q11.23_dup_distal	coagulation defects	378934	23	2541	0	0.2	0.00	0.49	3.24	0.03	23.30	1728	0.89
7q11.23_dup_distal	irritable bowel syndrome	368471	22	13004	1	0.8	1.28	0.49	1.89	0.21	7.38	1741	0.89
7q11.23_dup_distal	low WBC count	378510	23	2965	0	0.2	0.00	0.55	2.72	0.02	19.58	1877	0.91
7q11.23_dup_distal	nasal	378283	23	3192	0	0.2	0.00	0.56	2.60	0.02	18.73	1928	0.91
7q11.23_dup_distal	thyroid	354333	21	27142	2	1.6	1.22	0.56	1.51	0.30	4.90	1930	0.91
7q11.23_dup_distal	prostate hyperplasia	367027	22	14448	1	0.9	1.15	0.57	1.76	0.18	8.81	1933	0.92
7q11.23_dup_distal	sciatica	338361	20	43114	3	2.5	1.15	0.58	1.39	0.37	3.84	1968	0.93
7q11.23_dup_distal	ischaemic heart disease not MI	365152	23	16323	0	1.0	0.00	0.61	0.52	0.00	3.83	2033	0.94
7q11.23_dup_distal	migraine	367797	23	13678	0	0.9	0.00	0.62	0.53	0.00	3.83	2060	0.94
7q11.23_dup_distal	neuropathies	365571	22	15904	1	1.0	1.04	0.65	1.50	0.17	5.84	2155	0.95
7q11.23_dup_distal	cerebrovascular disease not stroke	377145	23	4330	0	0.3	0.00	0.66	2.04	0.02	15.06	2166	0.95
7q11.23_dup_distal	ear	367256	23	14219	0	0.9	0.00	0.67	0.58	0.00	4.16	2220	0.95
7q11.23_dup_distal	gastric reflux	347179	22	34296	1	2.2	0.48	0.68	0.72	0.08	2.78	2236	0.95
7q11.23_dup_distal	asthma	332149	21	49326	2	3.1	0.67	0.72	0.79	0.16	2.46	2330	0.96
7q11.23_dup_distal	heartfailure	376400	23	5075	0	0.3	0.00	0.72	1.77	0.01	13.33	2332	0.96
7q11.23_dup_distal	inflammatory bowel disease	362884	22	18591	1	1.1	0.89	0.73	1.36	0.15	5.28	2372	0.96
7q11.23_dup_distal	any Ca	331777	21	49698	2	3.1	0.67	0.74	0.80	0.16	2.55	2408	0.96
7q11.23_dup_distal	anaemia	362334	22	19141	1	1.2	0.87	0.75	1.33	0.15	5.16	2425	0.97
7q11.23_dup_distal	atherosclerotic vascular disease	375726	23	5749	0	0.4	0.00	0.77	1.55	0.01	11.36	2510	0.97
7q11.23_dup_distal	ureter bladder	332946	21	48529	2	3.1	0.68	0.78	0.83	0.17	2.61	2525	0.97
7q11.23_dup_distal	psoriasis	375455	23	6020	0	0.4	0.00	0.82	1.41	0.01	10.16	2642	0.97
7q11.23_dup_distal	biliary	360700	22	20775	1	1.3	0.80	0.84	1.19	0.13	4.65	2720	0.97
7q11.23_dup_distal	osteoarthritis	312766	19	68709	4	4.2	0.97	0.85	1.11	0.34	2.91	2737	0.97
7q11.23_dup_distal	cataract	359624	22	21851	1	1.3	0.76	0.87	1.16	0.13	4.76	2779	0.98
7q11.23_dup_distal	ocular	359814	22	21661	1	1.3	0.77	0.89	1.13	0.12	4.39	2847	0.98
7q11.23_dup_distal	gout	374385	23	7090	0	0.4	0.00	0.90	1.21	0.01	9.35	2870	0.98
7q11.23_dup_distal	obesity	371638	23	9837	0	0.6	0.00	0.91	0.85	0.01	6.13	2887	0.98
7q11.23_dup_distal	renal	374186	23	7289	0	0.4	0.00	0.92	1.15	0.01	8.29	2935	0.99
7q11.23_dup_distal	connective tissue disorder	372123	23	9352	0	0.6	0.00	0.92	0.87	0.01	6.38	2936	0.99
7q11.23_dup_distal	hypertension	260837	16	120638	7	7.4	0.96	0.92	1.04	0.40	2.49	2939	0.99
7q11.23_dup_distal	gastrointestinal ulcer	371123	23	10352	0	0.6	0.00	0.93	0.88	0.01	6.40	2943	0.98

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
8p23.1_dup	nasal	378264	4	3193	1	0.0	23.90	0.0071	34.71	3.42	194.16	96	0.23
8p23.1_dup	cerebrovascular_disease_not_stroke	377126	4	4331	1	0.0	17.62	0.012	25.97	2.52	148.08	134	0.27
8p23.1_dup	obesity	371619	4	9838	1	0.1	7.76	0.033	12.96	1.29	70.70	256	0.41
8p23.1_dup	irritable_bowel_syndrome	368453	4	13004	1	0.1	5.87	0.041	11.60	1.13	66.28	296	0.44
8p23.1_dup	COPD	367228	4	14229	1	0.2	5.36	0.051	9.97	0.99	54.52	331	0.48
8p23.1_dup	digestive	299610	2	81847	3	0.5	2.80	0.056	4.92	0.95	29.63	356	0.50
8p23.1_dup	ear	367237	4	14220	1	0.2	5.37	0.069	8.14	0.81	44.20	395	0.55
8p23.1_dup	hypertension	260826	5	120631	0	2.3	0.00	0.097	0.14	0.00	1.32	514	0.59
8p23.1_dup	inflammatory_bowel_disease	362866	4	18591	1	0.2	4.10	0.097	6.47	0.65	35.15	520	0.59
8p23.1_dup	any_Ca	331759	3	49698	2	0.4	3.07	0.11	4.19	0.68	22.07	565	0.63
8p23.1_dup	respiratory	360706	4	20751	1	0.2	3.68	0.13	5.47	0.55	29.83	620	0.64
8p23.1_dup	thyroid	354316	4	27141	1	0.3	2.81	0.16	5.05	0.46	32.24	715	0.69
8p23.1_dup	paralytic_syndromes	379497	5	1960	0	0.0	0.00	0.18	16.75	0.13	150.07	763	0.72
8p23.1_dup	endocrine_other	379097	5	2360	0	0.0	0.00	0.19	14.38	0.11	130.12	817	0.74
8p23.1_dup	coagulation_defects	378916	5	2541	0	0.0	0.00	0.20	13.44	0.10	119.18	847	0.74
8p23.1_dup	diabetes_insulin_dependent	378863	5	2594	0	0.0	0.00	0.22	12.12	0.09	108.77	890	0.76
8p23.1_dup	aneurism	379491	5	1966	0	0.0	0.00	0.22	12.25	0.09	119.98	892	0.76
8p23.1_dup	low_WBC_count	378492	5	2965	0	0.0	0.00	0.22	11.42	0.09	101.25	910	0.77
8p23.1_dup	congenital	376430	5	5027	0	0.1	0.00	0.31	6.84	0.05	60.46	1186	0.82
8p23.1_dup	atherosclerotic_vascular_disease	375708	5	5749	0	0.1	0.00	0.33	6.04	0.05	54.42	1270	0.83
8p23.1_dup	psoriasis	375437	5	6020	0	0.1	0.00	0.34	5.83	0.04	51.56	1294	0.83
8p23.1_dup	heartfailure	376382	5	5075	0	0.1	0.00	0.34	5.86	0.04	55.68	1302	0.83
8p23.1_dup	ovarian_cysts	374069	5	7388	0	0.1	0.00	0.35	5.95	0.04	73.34	1306	0.83
8p23.1_dup	high_cholesterol	314877	5	66580	0	1.1	0.00	0.39	0.33	0.00	3.05	1425	0.85
8p23.1_dup	asthma	332132	4	49325	1	0.6	1.55	0.39	2.41	0.24	13.06	1449	0.85
8p23.1_dup	renal	374168	5	7289	0	0.1	0.00	0.41	4.35	0.03	38.40	1497	0.86
8p23.1_dup	glaucoma	374929	5	6528	0	0.1	0.00	0.44	4.00	0.03	37.13	1568	0.87
8p23.1_dup	hepatic	372704	5	8753	0	0.1	0.00	0.44	3.94	0.03	34.86	1573	0.87
8p23.1_dup	connective_tissue_disorder	372105	5	9352	0	0.1	0.00	0.45	3.74	0.03	34.00	1624	0.88
8p23.1_dup	renal_failure	373537	5	7920	0	0.1	0.00	0.46	3.63	0.03	33.08	1647	0.88
8p23.1_dup	stroke_derived2017	372639	5	8818	0	0.1	0.00	0.48	3.41	0.03	30.85	1709	0.88
8p23.1_dup	heart_valve	373081	5	8376	0	0.1	0.00	0.48	3.40	0.03	30.60	1710	0.88
8p23.1_dup	gout	374367	5	7090	0	0.1	0.00	0.49	3.41	0.03	34.87	1721	0.88
8p23.1_dup	gastrointestinal_ulcer	371105	5	10352	0	0.1	0.00	0.49	3.30	0.03	29.57	1734	0.89
8p23.1_dup	osteoporosis	369864	5	11593	0	0.2	0.00	0.51	3.14	0.02	32.24	1793	0.90
8p23.1_dup	varicose_veins	369605	5	11852	0	0.2	0.00	0.52	3.01	0.02	26.72	1817	0.90
8p23.1_dup	died_to_2016	370488	5	10969	0	0.1	0.00	0.53	2.94	0.02	26.43	1836	0.90
8p23.1_dup	migraine	367779	5	13678	0	0.2	0.00	0.53	2.91	0.02	27.26	1849	0.90
8p23.1_dup	heart_other	369636	5	11821	0	0.2	0.00	0.62	2.25	0.02	20.81	2072	0.94
8p23.1_dup	hernia	333569	5	47888	0	0.7	0.00	0.62	0.51	0.00	4.68	2076	0.94
8p23.1_dup	venous_thromboembolic_disease	367485	5	13972	0	0.2	0.00	0.63	2.20	0.02	19.56	2101	0.94
8p23.1_dup	allergy	322797	5	58660	0	0.9	0.00	0.63	0.52	0.00	4.67	2102	0.94
8p23.1_dup	neuropathies	365554	5	15903	0	0.2	0.00	0.65	2.08	0.02	18.70	2161	0.95
8p23.1_dup	ureter_bladder	332930	5	48527	0	0.7	0.00	0.66	0.55	0.00	4.92	2186	0.95
8p23.1_dup	MI_derived2017	367597	5	13860	0	0.2	0.00	0.69	1.92	0.01	18.84	2271	0.95
8p23.1_dup	anaemia	362317	5	19140	0	0.3	0.00	0.72	1.76	0.01	15.71	2367	0.96
8p23.1_dup	osteoarthritis	312751	4	68706	1	0.9	1.11	0.73	1.40	0.14	7.78	2382	0.96
8p23.1_dup	ischaemic_heart_disease_not_MI	365134	5	16323	0	0.2	0.00	0.75	1.68	0.01	15.62	2421	0.97
8p23.1_dup	biliary	360683	5	20774	0	0.3	0.00	0.76	1.62	0.01	14.75	2466	0.97
8p23.1_dup	sciatica	338346	5	43111	0	0.6	0.00	0.83	0.73	0.01	6.46	2655	0.97
8p23.1_dup	diabetes_other	360723	5	20734	0	0.3	0.00	0.84	1.36	0.01	12.41	2714	0.97
8p23.1_dup	ocular	359797	5	21660	0	0.3	0.00	0.86	1.32	0.01	11.77	2761	0.97
8p23.1_dup	cataract	359607	5	21850	0	0.3	0.00	0.89	1.24	0.01	11.77	2843	0.98
8p23.1_dup	uterine_problem	339169	5	42288	0	0.6	0.00	0.89	0.81	0.01	10.03	2851	0.98
8p23.1_dup	prostate_hyperplasia	367010	5	14447	0	0.2	0.00	0.94	1.12	0.01	13.22	2991	0.99
8p23.1_dup	gastric_reflux	347162	5	34295	0	0.5	0.00	0.96	0.93	0.01	8.20	3031	0.99
8p23.1_dup	arrhythmia	357299	5	24158	0	0.3	0.00	0.97	1.06	0.01	9.77	3058	0.99
8p23.1_dup	diverticular_disease_intestine	354324	5	27133	0	0.4	0.00	0.97	1.05	0.01	9.59	3072	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
NRXN1_del	aneurism	379638	152	1971	5	0.8	6.18	0.00042	7.59	2.82	16.42	32	0.041
NRXN1_del	digestive	299718	110	81891	47	30.1	1.40	0.0065	1.64	1.15	2.30	91	0.22
NRXN1_del	venous_thromboembolic_disease	367626	146	13983	11	5.6	1.91	0.027	2.13	1.10	3.72	227	0.37
NRXN1_del	atherosclerotic_vascular_disease	375855	152	5754	5	2.3	2.11	0.073	2.44	0.91	5.23	419	0.55
NRXN1_del	asthma	332258	130	49351	27	19.3	1.33	0.11	1.42	0.92	2.10	556	0.63
NRXN1_del	heart_other	369780	149	11829	8	4.8	1.64	0.11	1.86	0.85	3.51	561	0.63
NRXN1_del	thyroid	354454	142	27155	15	10.9	1.34	0.13	1.55	0.87	2.57	644	0.64
NRXN1_del	obesity	371765	150	9844	7	4.0	1.73	0.14	1.84	0.80	3.58	664	0.65
NRXN1_del	stroke_derived2017	372785	151	8824	6	3.6	1.65	0.16	1.88	0.76	3.83	707	0.69
NRXN1_del	diabetes_other	360863	145	20746	12	8.3	1.41	0.18	1.54	0.81	2.65	766	0.72
NRXN1_del	hepatic	372855	156	8754	1	3.7	0.28	0.20	0.41	0.05	1.46	831	0.75
NRXN1_del	hypertension	260923	102	120686	55	47.2	1.11	0.21	1.25	0.88	1.75	877	0.75
NRXN1_del	arrhythmia	357438	144	24171	13	9.7	1.31	0.22	1.45	0.79	2.46	896	0.76
NRXN1_del	diverticular_disease_intestine	354462	143	27147	14	11.0	1.25	0.22	1.43	0.79	2.40	899	0.76
NRXN1_del	renal_failure	373684	152	7925	5	3.2	1.53	0.23	1.76	0.66	3.78	945	0.78
NRXN1_del	ocular	359944	152	21665	5	9.1	0.56	0.24	0.62	0.23	1.32	950	0.78
NRXN1_del	hernia	333697	133	47912	24	19.1	1.22	0.25	1.31	0.82	1.99	985	0.78
NRXN1_del	diabetes_insulin_dependent	379013	155	2596	2	1.1	1.87	0.25	2.28	0.48	6.50	1012	0.79
NRXN1_del	ischaemic_heart_disease_not_MI	365277	148	16332	9	6.6	1.34	0.26	1.50	0.72	2.78	1030	0.79
NRXN1_del	glaucoma	375077	153	6532	4	2.7	1.49	0.27	1.79	0.59	4.09	1047	0.80
NRXN1_del	heart_valve	373228	152	8381	5	3.4	1.45	0.28	1.67	0.62	3.56	1078	0.80
NRXN1_del	ear	367387	154	14222	3	6.0	0.51	0.28	0.59	0.16	1.46	1114	0.80
NRXN1_del	biliary	360830	152	20779	5	8.8	0.58	0.31	0.66	0.25	1.40	1180	0.81
NRXN1_del	MI_derived2017	367746	154	13863	3	5.8	0.53	0.31	0.60	0.17	1.52	1202	0.82
NRXN1_del	ovarian_cysts	374220	156	7389	1	3.1	0.33	0.33	0.49	0.06	1.77	1241	0.82
NRXN1_del	connective_tissue_disorder	372252	152	9357	5	3.8	1.30	0.38	1.51	0.56	3.22	1387	0.85
NRXN1_del	neuropathies	365702	153	15907	4	6.7	0.61	0.39	0.68	0.23	1.54	1433	0.85
NRXN1_del	renal	374316	153	7293	4	3.0	1.33	0.40	1.54	0.51	3.49	1478	0.85
NRXN1_del	COPD	367374	150	14235	7	5.8	1.20	0.42	1.38	0.60	2.71	1523	0.87
NRXN1_del	allergy	322928	136	58681	21	24.7	0.87	0.47	0.85	0.52	1.31	1672	0.88
NRXN1_del	prostate_hyperplasia	367158	153	14451	4	6.0	0.67	0.48	0.71	0.23	1.70	1694	0.88
NRXN1_del	psoriasis	375588	156	6021	1	2.5	0.40	0.48	0.59	0.07	2.12	1705	0.88
NRXN1_del	coagulation_defects	379068	157	2541	0	1.1	0.00	0.54	0.47	0.00	3.21	1869	0.91
NRXN1_del	migraine	367927	153	13682	4	5.7	0.71	0.57	0.77	0.26	1.75	1938	0.92
NRXN1_del	endocrine_other	379248	156	2361	1	1.0	1.03	0.60	1.58	0.18	5.68	2021	0.94
NRXN1_del	anaemia	362463	151	19146	6	8.0	0.76	0.62	0.82	0.34	1.67	2062	0.94
NRXN1_del	osteoarthritis	312875	128	68734	29	28.1	1.03	0.63	1.11	0.72	1.65	2089	0.94
NRXN1_del	died_to_2016	370635	152	10974	5	4.5	1.11	0.67	1.21	0.45	2.59	2192	0.95
NRXN1_del	osteoporosis	370013	154	11596	3	4.8	0.63	0.67	0.80	0.22	2.02	2214	0.95
NRXN1_del	heartfailure	376533	156	5076	1	2.1	0.48	0.68	0.72	0.08	2.62	2223	0.95
NRXN1_del	paralytic_syndromes	379649	157	1960	0	0.8	0.00	0.68	0.59	0.00	4.02	2244	0.95
NRXN1_del	sciatica	338479	138	43130	19	17.6	1.07	0.69	1.10	0.66	1.73	2272	0.95
NRXN1_del	respiratory	360852	150	20757	7	8.6	0.82	0.72	0.87	0.38	1.70	2331	0.96
NRXN1_del	cataract	359752	150	21857	7	9.1	0.78	0.74	0.88	0.38	1.74	2405	0.96
NRXN1_del	varicose_veins	369752	152	11857	5	4.9	1.02	0.74	1.16	0.43	2.46	2409	0.96
NRXN1_del	inflammatory_bowel_disease	363011	149	18598	8	7.6	1.05	0.74	1.13	0.52	2.11	2412	0.96
NRXN1_del	low_WBC_count	378643	156	2966	1	1.2	0.82	0.77	1.28	0.15	4.60	2500	0.97
NRXN1_del	gout	374517	155	7092	2	2.9	0.69	0.78	0.84	0.17	2.41	2512	0.97
NRXN1_del	congenital	376580	155	5029	2	2.1	0.97	0.79	1.19	0.25	3.38	2557	0.97
NRXN1_del	ureter_bladder	333062	137	48547	20	20.0	1.00	0.82	1.06	0.64	1.65	2636	0.97
NRXN1_del	high_cholesterol	315002	130	66607	27	27.5	0.99	0.85	1.04	0.67	1.58	2734	0.97
NRXN1_del	cerebrovascular_disease_not_stroke	377278	156	4331	1	1.8	0.56	0.87	0.88	0.10	3.17	2781	0.98
NRXN1_del	gastrointestinal_ulcer	371253	153	10356	4	4.3	0.94	0.87	1.08	0.36	2.47	2785	0.98
NRXN1_del	gastric_reflux	347300	143	34309	14	14.1	0.99	0.88	1.04	0.58	1.73	2807	0.98
NRXN1_del	nasal	378416	156	3193	1	1.3	0.76	0.88	1.13	0.13	4.07	2818	0.98
NRXN1_del	uterine_problem	339305	141	42304	16	17.6	0.92	0.91	0.97	0.55	1.63	2899	0.98
NRXN1_del	irritable_bowel_syndrome	368601	152	13008	5	5.4	0.93	0.94	1.03	0.39	2.20	2990	0.99
NRXN1_del	any_Ca	331894	138	49715	19	20.7	0.93	0.98	1.01	0.60	1.59	3085	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
Potocki_Lupski	diabetes_insulin_dependent	378862	4	2595	1	0.0	29.41	0.0041	48.85	4.83	270.84	74	0.17
Potocki_Lupski	thyroid	354315	3	27142	2	0.2	5.62	0.022	9.47	1.44	55.75	201	0.34
Potocki_Lupski	heart_valve	373080	4	8377	1	0.1	9.11	0.027	14.81	1.46	82.01	232	0.37
Potocki_Lupski	hepatic	372703	4	8754	1	0.1	8.72	0.034	12.84	1.28	70.55	261	0.41
Potocki_Lupski	connective_tissue_disorder	372104	4	9353	1	0.1	8.16	0.037	12.19	1.21	67.29	275	0.42
Potocki_Lupski	osteoporosis	369863	4	11594	1	0.1	6.58	0.043	11.56	1.10	70.25	303	0.45
Potocki_Lupski	died_to_2016	370487	4	10970	1	0.1	6.96	0.048	10.52	1.02	60.19	322	0.47
Potocki_Lupski	migraine	367778	4	13679	1	0.1	5.58	0.063	8.90	0.86	52.19	373	0.53
Potocki_Lupski	ear	367237	4	14220	1	0.2	5.37	0.065	8.41	0.84	45.59	380	0.54
Potocki_Lupski	ureter_bladder	332928	3	48529	2	0.4	3.14	0.085	4.77	0.78	24.94	470	0.57
Potocki_Lupski	anaemia	362316	4	19141	1	0.2	3.99	0.12	5.72	0.57	31.09	586	0.63
Potocki_Lupski	aneurism	379491	5	1966	0	0.0	0.00	0.18	16.20	0.12	174.61	789	0.73
Potocki_Lupski	paralytic_syndromes	379497	5	1960	0	0.0	0.00	0.19	15.36	0.12	139.87	799	0.73
Potocki_Lupski	endocrine_other	379097	5	2360	0	0.0	0.00	0.20	13.31	0.10	119.54	855	0.74
Potocki_Lupski	coagulation_defects	378916	5	2541	0	0.0	0.00	0.21	13.00	0.10	115.16	864	0.74
Potocki_Lupski	nasal	378265	5	3192	0	0.0	0.00	0.22	11.89	0.09	106.96	893	0.76
Potocki_Lupski	low_WBC_count	378492	5	2965	0	0.0	0.00	0.22	11.36	0.09	100.84	913	0.77
Potocki_Lupski	cerebrovascular_disease_not_stroke	377127	5	4330	0	0.1	0.00	0.28	8.28	0.06	77.75	1077	0.80
Potocki_Lupski	digestive	299611	3	81846	2	0.8	1.86	0.29	2.49	0.41	12.88	1135	0.81
Potocki_Lupski	congenital	376430	5	5027	0	0.1	0.00	0.32	6.32	0.05	55.90	1239	0.82
Potocki_Lupski	heartfailure	376382	5	5075	0	0.1	0.00	0.34	6.10	0.04	62.82	1286	0.82
Potocki_Lupski	atherosclerotic_vascular_disease	375708	5	5749	0	0.1	0.00	0.35	5.55	0.04	51.87	1328	0.84
Potocki_Lupski	glaucoma	374929	5	6528	0	0.1	0.00	0.37	5.25	0.04	49.49	1363	0.85
Potocki_Lupski	psoriasis	375437	5	6020	0	0.1	0.00	0.37	5.16	0.04	46.17	1369	0.85
Potocki_Lupski	renal	374168	5	7289	0	0.1	0.00	0.40	4.48	0.03	39.68	1482	0.85
Potocki_Lupski	gout	374367	5	7090	0	0.1	0.00	0.42	4.39	0.03	52.25	1515	0.87
Potocki_Lupski	asthma	332132	4	49325	1	0.6	1.55	0.42	2.26	0.23	12.30	1528	0.87
Potocki_Lupski	uterine_problem	339168	4	42289	1	0.5	1.80	0.43	2.36	0.22	17.74	1543	0.87
Potocki_Lupski	any_Ca	331760	4	49697	1	0.6	1.54	0.44	2.21	0.22	12.40	1580	0.87
Potocki_Lupski	renal_failure	373537	5	7920	0	0.1	0.00	0.44	3.94	0.03	37.28	1584	0.87
Potocki_Lupski	ovarian_cysts	374069	5	7388	0	0.1	0.00	0.44	3.96	0.03	40.93	1585	0.87
Potocki_Lupski	osteoarthritis	312752	5	68705	0	1.1	0.00	0.47	0.39	0.00	3.52	1679	0.88
Potocki_Lupski	stroke_derived2017	372639	5	8818	0	0.1	0.00	0.48	3.43	0.03	32.26	1711	0.88
Potocki_Lupski	allergy	322796	4	58661	1	0.7	1.30	0.50	1.97	0.20	10.79	1767	0.89
Potocki_Lupski	obesity	371620	5	9837	0	0.1	0.00	0.51	3.08	0.02	27.58	1797	0.89
Potocki_Lupski	gastrointestinal_ulcer	371105	5	10352	0	0.1	0.00	0.52	3.05	0.02	28.22	1809	0.90
Potocki_Lupski	heart_other	369636	5	11821	0	0.2	0.00	0.56	2.72	0.02	25.87	1911	0.91
Potocki_Lupski	varicose_veins	369605	5	11852	0	0.2	0.00	0.57	2.60	0.02	22.99	1942	0.92
Potocki_Lupski	COPD	367229	5	14228	0	0.2	0.00	0.59	2.49	0.02	22.84	1976	0.93
Potocki_Lupski	irritable_bowel_syndrome	368454	5	13003	0	0.2	0.00	0.59	2.43	0.02	22.23	1999	0.93
Potocki_Lupski	venous_thromboembolic_disease	367485	5	13972	0	0.2	0.00	0.62	2.27	0.02	20.23	2061	0.94
Potocki_Lupski	high_cholesterol	314876	4	66581	1	0.8	1.15	0.64	1.61	0.15	9.44	2128	0.95
Potocki_Lupski	hypertension	260824	3	120633	2	1.4	1.26	0.65	1.49	0.24	8.03	2144	0.95
Potocki_Lupski	MI_derived2017	367597	5	13860	0	0.2	0.00	0.67	2.06	0.01	23.10	2202	0.95
Potocki_Lupski	neuropathies	365554	5	15903	0	0.2	0.00	0.68	1.96	0.01	17.64	2225	0.95
Potocki_Lupski	ischaemic_heart_disease_not_MI	365134	5	16323	0	0.2	0.00	0.69	1.95	0.01	18.88	2255	0.95
Potocki_Lupski	hernia	333569	5	47888	0	0.7	0.00	0.73	0.62	0.00	5.70	2377	0.96
Potocki_Lupski	prostate_hyperplasia	367010	5	14447	0	0.2	0.00	0.75	1.73	0.01	28.01	2423	0.97
Potocki_Lupski	inflammatory_bowel_disease	362867	5	18590	0	0.3	0.00	0.76	1.64	0.01	14.48	2452	0.97
Potocki_Lupski	sciatica	338346	5	43111	0	0.6	0.00	0.79	0.69	0.01	6.09	2559	0.97
Potocki_Lupski	diabetes_other	360723	5	20734	0	0.3	0.00	0.80	1.50	0.01	14.11	2580	0.97
Potocki_Lupski	ocular	359797	5	21660	0	0.3	0.00	0.80	1.49	0.01	13.41	2583	0.97
Potocki_Lupski	respiratory	360707	5	20750	0	0.3	0.00	0.81	1.45	0.01	13.11	2619	0.97
Potocki_Lupski	biliary	360683	5	20774	0	0.3	0.00	0.81	1.44	0.01	12.96	2626	0.97
Potocki_Lupski	cataract	359607	5	21850	0	0.3	0.00	0.82	1.44	0.01	14.22	2630	0.97
Potocki_Lupski	arrhythmia	357299	5	24158	0	0.3	0.00	0.87	1.28	0.01	11.99	2791	0.98
Potocki_Lupski	diverticular_disease_intestine	354324	5	27133	0	0.4	0.00	0.91	1.20	0.01	10.92	2888	0.98
Potocki_Lupski	gastric_reflux	347162	5	34295	0	0.5	0.00	0.95	0.92	0.01	8.13	3024	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
PWS_dup	COPD	367237	13	14231	3	0.5	5.03	0.0018	11.84	2.90	37.33	51	0.11
PWS_dup	glaucoma	374938	14	6530	2	0.2	7.30	0.0033	16.99	3.09	62.56	67	0.15
PWS_dup	irritable_bowel_syndrome	368462	13	13006	3	0.5	5.50	0.011	6.55	1.66	19.70	128	0.26
PWS_dup	arrhythmia	357307	13	24161	3	0.9	2.96	0.014	6.31	1.54	19.86	148	0.29
PWS_dup	paralytic_syndromes	379507	15	1961	1	0.1	12.16	0.015	19.39	2.11	79.69	158	0.30
PWS_dup	hepatic	372713	14	8755	2	0.3	5.45	0.018	7.79	1.53	25.71	173	0.33
PWS_dup	coagulation_defects	378926	15	2542	1	0.1	9.38	0.022	15.33	1.68	61.92	199	0.34
PWS_dup	cerebrovascular_disease_not_stroke	377137	15	4331	1	0.2	5.51	0.027	14.24	1.48	64.03	229	0.37
PWS_dup	neuropathies	365563	14	15905	2	0.6	3.00	0.076	4.23	0.83	13.94	433	0.55
PWS_dup	ocular	359806	14	21662	2	0.8	2.20	0.093	3.92	0.76	13.22	500	0.58
PWS_dup	stroke_derived2017	372649	15	8819	1	0.4	2.70	0.094	6.20	0.66	26.40	506	0.58
PWS_dup	renal	374178	15	7290	1	0.3	3.27	0.11	5.56	0.61	22.46	546	0.62
PWS_dup	biliary	360692	14	20776	2	0.8	2.30	0.11	3.61	0.71	12.00	550	0.62
PWS_dup	inflammatory_bowel_disease	362876	14	18592	2	0.7	2.56	0.12	3.48	0.68	11.43	585	0.63
PWS_dup	anaemia	362326	14	19142	2	0.7	2.49	0.13	3.34	0.66	11.03	629	0.64
PWS_dup	osteoporosis	369874	15	11594	1	0.5	2.06	0.14	5.08	0.51	24.00	662	0.65
PWS_dup	MI_derived2017	367607	15	13861	1	0.6	1.72	0.17	4.42	0.45	20.57	738	0.70
PWS_dup	asthma	332140	12	49328	4	1.8	1.93	0.18	2.20	0.66	6.07	786	0.73
PWS_dup	died_to_2016	370498	15	10970	1	0.4	2.17	0.19	3.87	0.42	15.85	801	0.73
PWS_dup	ureter_bladder	332938	13	48530	3	1.9	1.47	0.19	2.37	0.60	7.12	819	0.74
PWS_dup	sciatica	338357	16	43111	0	2.0	0.00	0.24	0.26	0.00	1.91	955	0.78
PWS_dup	aneurism	379502	16	1966	0	0.1	0.00	0.25	9.47	0.07	79.51	1005	0.78
PWS_dup	any_Ca	331772	16	49696	0	2.4	0.00	0.30	0.29	0.00	2.26	1161	0.81
PWS_dup	cataract	359617	15	21851	1	0.9	1.09	0.33	2.78	0.28	14.05	1244	0.82
PWS_dup	endocrine_other	379108	16	2360	0	0.1	0.00	0.37	5.13	0.04	38.19	1355	0.84
PWS_dup	digestive	299620	12	81848	4	3.3	1.17	0.38	1.67	0.50	4.68	1393	0.85
PWS_dup	diabetes_insulin_dependent	378874	16	2594	0	0.1	0.00	0.39	4.59	0.04	34.45	1454	0.85
PWS_dup	nasal	378276	16	3192	0	0.1	0.00	0.40	4.47	0.03	33.70	1472	0.85
PWS_dup	low_WBC_count	378503	16	2965	0	0.1	0.00	0.41	4.28	0.03	31.73	1495	0.86
PWS_dup	gastric_reflux	347173	16	34295	0	1.6	0.00	0.44	0.38	0.00	2.86	1565	0.87
PWS_dup	uterine_problem	339177	13	42291	3	1.6	1.69	0.46	1.65	0.40	5.55	1631	0.88
PWS_dup	diverticular_disease_intestine	354334	15	27134	1	1.1	0.88	0.46	2.05	0.22	8.90	1638	0.88
PWS_dup	heartfailure	376393	16	5075	0	0.2	0.00	0.47	3.52	0.03	29.04	1665	0.88
PWS_dup	thyroid	354328	16	27140	0	1.2	0.00	0.51	0.43	0.00	3.31	1777	0.89
PWS_dup	atherosclerotic_vascular_disease	375719	16	5749	0	0.2	0.00	0.53	2.91	0.02	22.43	1831	0.90
PWS_dup	osteoarthritis	312762	15	68706	1	3.3	0.35	0.60	0.65	0.07	2.76	2014	0.94
PWS_dup	gout	374378	16	7090	0	0.3	0.00	0.63	2.23	0.02	18.68	2080	0.94
PWS_dup	congenital	376441	16	5027	0	0.2	0.00	0.66	2.03	0.02	15.03	2171	0.95
PWS_dup	renal_failure	373548	16	7920	0	0.3	0.00	0.66	2.04	0.02	16.10	2173	0.95
PWS_dup	high_cholesterol	314887	15	66581	1	3.2	0.36	0.68	0.70	0.07	3.15	2228	0.95
PWS_dup	heart_valve	373092	16	8376	0	0.4	0.00	0.68	1.91	0.01	14.58	2251	0.95
PWS_dup	psoriasis	375448	16	6020	0	0.3	0.00	0.68	1.90	0.01	14.06	2254	0.95
PWS_dup	migraine	367790	16	13678	0	0.6	0.00	0.72	0.62	0.00	4.66	2342	0.96
PWS_dup	respiratory	360718	16	20750	0	0.9	0.00	0.75	0.65	0.01	4.86	2420	0.97
PWS_dup	gastrointestinal_ulcer	371116	16	10352	0	0.4	0.00	0.78	1.53	0.01	11.62	2531	0.97
PWS_dup	connective_tissue_disorder	372116	16	9352	0	0.4	0.00	0.78	1.53	0.01	11.51	2539	0.97
PWS_dup	hypertension	260834	13	120634	3	6.0	0.59	0.79	0.85	0.21	2.69	2562	0.97
PWS_dup	diabetes_other	360734	16	20734	0	0.9	0.00	0.80	0.71	0.01	5.52	2596	0.97
PWS_dup	heart_other	369647	16	11821	0	0.5	0.00	0.85	1.33	0.01	10.49	2739	0.97
PWS_dup	allergy	322806	14	58662	2	2.5	0.81	0.88	0.90	0.18	2.95	2819	0.98
PWS_dup	hernia	333579	15	47889	1	2.2	0.50	0.92	0.91	0.10	3.84	2916	0.98
PWS_dup	prostate_hyperplasia	367021	16	14447	0	0.6	0.00	0.94	0.90	0.01	10.68	2997	0.99
PWS_dup	obesity	371631	16	9837	0	0.4	0.00	0.95	1.10	0.01	8.20	3006	0.99
PWS_dup	ear	367249	16	14219	0	0.6	0.00	0.97	0.95	0.01	7.04	3061	0.99
PWS_dup	ischaemic_heart_disease_not_MI	365145	16	16323	0	0.7	0.00	0.98	1.04	0.01	8.50	3080	0.99
PWS_dup	varicose_veins	369616	16	11852	0	0.5	0.00	0.98	1.04	0.01	7.72	3082	0.99
PWS_dup	venous_thromboembolic_disease	367496	16	13972	0	0.6	0.00	0.99	0.98	0.01	7.33	3103	1.00
PWS_dup	ovarian_cysts	374080	16	7388	0	0.3	0.00	0.99	0.99	0.01	7.72	3119	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
TAR_del	COPD	367286	62	14235	7	2.4	2.72	0.0064	3.54	1.48	7.34	90	0.22
TAR_del	thyroid	354371	59	27150	10	4.5	2.04	0.016	2.49	1.20	4.70	166	0.31
TAR_del	respiratory	360763	61	20758	8	3.5	2.13	0.024	2.54	1.14	4.98	212	0.36
TAR_del	ocular	359861	69	21660	0	4.2	0.00	0.028	0.12	0.00	0.85	236	0.37
TAR_del	asthma	332194	66	49327	3	9.8	0.34	0.028	0.36	0.10	0.91	238	0.37
TAR_del	MI_derived2017	367655	63	13866	6	2.4	2.39	0.030	2.89	1.12	6.31	243	0.38
TAR_del	ureter_bladder	332980	55	48541	14	8.0	1.59	0.043	1.90	1.02	3.33	302	0.45
TAR_del	inflammatory_bowel_disease	362931	69	18590	0	3.5	0.00	0.048	0.14	0.00	0.99	323	0.47
TAR_del	hypertension	260862	41	120659	28	19.0	1.28	0.053	1.67	0.99	2.75	339	0.49
TAR_del	hepatic	372764	65	8757	4	1.5	2.53	0.053	3.00	0.98	7.01	342	0.49
TAR_del	glaucoma	374990	66	6531	3	1.1	2.54	0.076	3.17	0.87	8.19	432	0.55
TAR_del	cerebrovascular_disease_not_stroke	377189	67	4332	2	0.8	2.55	0.11	3.42	0.70	10.08	555	0.63
TAR_del	ovarian_cysts	374130	66	7391	3	1.3	2.24	0.13	2.62	0.71	6.89	639	0.64
TAR_del	renal_failure	373598	66	7923	3	1.4	2.09	0.13	2.58	0.70	6.68	653	0.65
TAR_del	aneurism	379554	68	1967	1	0.4	2.81	0.16	4.24	0.48	15.91	716	0.69
TAR_del	atherosclerotic_vascular_disease	375770	67	5751	2	1.0	1.92	0.21	2.53	0.52	7.43	882	0.75
TAR_del	venous_thromboembolic_disease	367545	65	13976	4	2.5	1.58	0.23	1.89	0.62	4.42	939	0.78
TAR_del	obesity	371681	66	9840	3	1.7	1.69	0.23	2.07	0.57	5.28	940	0.78
TAR_del	low_WBC_count	378555	68	2966	1	0.5	1.86	0.28	2.84	0.32	10.38	1086	0.80
TAR_del	arrhythmia	357357	63	24164	6	4.3	1.37	0.29	1.60	0.64	3.39	1133	0.81
TAR_del	died_to_2016	370549	66	10972	3	2.0	1.51	0.31	1.84	0.51	4.71	1196	0.82
TAR_del	any_Ca	331814	58	49707	11	8.7	1.22	0.31	1.41	0.70	2.59	1207	0.81
TAR_del	prostate_hyperplasia	367070	65	14451	4	2.6	1.53	0.33	1.73	0.53	4.48	1252	0.82
TAR_del	ischaemic_heart_disease_not_MI	365197	68	16324	1	3.0	0.34	0.34	0.49	0.06	1.84	1301	0.83
TAR_del	sciatica	338405	64	43116	5	8.2	0.64	0.37	0.69	0.25	1.50	1372	0.85
TAR_del	varicose_veins	369666	66	11855	3	2.1	1.40	0.37	1.70	0.47	4.31	1380	0.85
TAR_del	gout	374431	69	7090	0	1.3	0.00	0.41	0.37	0.00	2.62	1493	0.86
TAR_del	renal	374232	69	7289	0	1.3	0.00	0.42	0.38	0.00	2.63	1522	0.87
TAR_del	heart_valve	373143	67	8378	2	1.5	1.32	0.43	1.74	0.36	5.09	1545	0.87
TAR_del	osteoarthritis	312807	60	68714	9	13.2	0.72	0.43	0.76	0.36	1.46	1554	0.87
TAR_del	cataract	359669	67	21852	2	4.1	0.51	0.49	0.65	0.13	1.94	1725	0.88
TAR_del	connective_tissue_disorder	372167	67	9354	2	1.7	1.18	0.50	1.59	0.33	4.65	1762	0.89</

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
TAR_dup	obesity	372000	385	9860	23	10.2	2.19	0.00054	2.29	1.47	3.40	36	0.047
TAR_dup	COPD	367606	382	14254	26	14.8	1.71	0.0056	1.86	1.21	2.73	83	0.21
TAR_dup	cataract	359975	373	21885	35	22.7	1.50	0.0058	1.71	1.18	2.40	84	0.22
TAR_dup	allergy	323119	327	58741	81	59.4	1.29	0.0092	1.40	1.09	1.77	118	0.25
TAR_dup	digestive	299909	301	81951	107	82.2	1.22	0.013	1.34	1.07	1.67	139	0.28
TAR_dup	hernia	333904	340	47956	68	48.8	1.33	0.020	1.39	1.05	1.80	185	0.34
TAR_dup	venous_thromboembolic_disease	367865	385	13995	23	14.6	1.54	0.031	1.64	1.05	2.43	249	0.39
TAR_dup	hypertension	261085	264	120775	144	122.1	1.12	0.067	1.22	0.99	1.51	385	0.54
TAR_dup	diabetes_other	361095	377	20765	31	21.7	1.40	0.068	1.44	0.97	2.04	389	0.55
TAR_dup	diabetes_insulin_dependent	379260	402	2600	6	2.8	2.16	0.073	2.23	0.92	4.46	420	0.55
TAR_dup	sciatica	338714	373	43146	35	47.5	0.76	0.096	0.75	0.52	1.05	511	0.59
TAR_dup	biliary	361058	380	20802	28	21.9	1.26	0.12	1.37	0.92	1.98	600	0.63
TAR_dup	any_Ca	332123	367	49737	41	55.0	0.77	0.13	0.78	0.56	1.07	619	0.63
TAR_dup	hepatic	373093	394	8767	14	9.3	1.50	0.13	1.54	0.87	2.50	650	0.64
TAR_dup	neuropathies	365935	386	15925	22	16.8	1.29	0.17	1.37	0.87	2.05	742	0.71
TAR_dup	MI_derived2017	367980	388	13880	20	14.6	1.35	0.18	1.38	0.85	2.12	787	0.73
TAR_dup	diverticular_disease_intestine	354693	374	27167	34	28.6	1.17	0.19	1.28	0.88	1.79	805	0.73
TAR_dup	gastrointestinal_ulcer	371493	393	10367	15	11.0	1.35	0.20	1.42	0.82	2.29	844	0.74
TAR_dup	gout	374759	397	7101	11	7.5	1.45	0.25	1.45	0.75	2.50	995	0.78
TAR_dup	psoriasis	375831	399	6029	9	6.4	1.40	0.27	1.46	0.71	2.63	1070	0.80
TAR_dup	irritable_bowel_syndrome	368840	391	13020	17	13.8	1.22	0.28	1.32	0.78	2.07	1093	0.80
TAR_dup	ocular	360173	381	21687	27	22.9	1.17	0.31	1.23	0.82	1.78	1187	0.81
TAR_dup	arrhythmia	357672	378	24188	30	25.6	1.16	0.32	1.22	0.82	1.74	1217	0.82
TAR_dup	prostate_hyperplasia	367401	396	14459	12	15.6	0.78	0.34	0.76	0.40	1.31	1292	0.83
TAR_dup	congenital	376830	405	5030	3	5.4	0.56	0.37	0.64	0.18	1.57	1360	0.85
TAR_dup	asthma	332478	350	49382	58	52.0	1.10	0.42	1.12	0.84	1.47	1506	0.87
TAR_dup	paralytic_syndromes	379897	405	1963	3	2.1	1.43	0.42	1.59	0.45	3.93	1521	0.87
TAR_dup	ovarian_cysts	374463	399	7397	9	7.9	1.14	0.43	1.32	0.64	2.39	1537	0.87
TAR_dup	high_cholesterol	315204	332	66656	76	70.2	1.07	0.43	1.11	0.85	1.43	1548	0.87
TAR_dup	osteoarthritis	313091	344	68769	64	75.6	0.87	0.43	0.90	0.68	1.17	1550	0.87
TAR_dup	heart_other	370024	393	11836	15	12.6	1.19	0.44	1.23	0.71	1.98	1582	0.87
TAR_dup	low_WBC_count	378891	404	2969	4	3.2	1.26	0.45	1.46	0.49	3.27	1623	0.88
TAR_dup	uterine_problem	339527	363	42333	45	45.3	0.99	0.45	1.14	0.81	1.57	1625	0.88
TAR_dup	endocrine_other	379499	407	2361	1	2.5	0.40	0.50	0.61	0.07	2.16	1772	0.89
TAR_dup	renal_failure	373930	398	7930	10	8.4	1.18	0.51	1.24	0.62	2.17	1800	0.89
TAR_dup	varicose_veins	369994	394	11866	14	12.6	1.10	0.52	1.20	0.67	1.94	1805	0.90
TAR_dup	ear	367624	391	14236	17	15.1	1.12	0.52	1.18	0.70	1.84	1818	0.90
TAR_dup	anaemia	362703	391	19157	17	20.7	0.83	0.54	0.86	0.51	1.35	1862	0.91
TAR_dup	atherosclerotic_vascular_disease	376104	401	5756	7	6.1	1.14	0.61	1.22	0.54	2.34	2026	0.94
TAR_dup	glaucoma	375327	403	6533	5	7.0	0.72	0.61	0.81	0.30	1.70	2037	0.94
TAR_dup	thyroid	354691	379	27169	29	29.0	1.00	0.63	1.10	0.74	1.58	2090	0.94
TAR_dup	gastric_reflux	347526	369	34334	39	36.5	1.06	0.66	1.08	0.76	1.49	2172	0.95
TAR_dup	coagulation_defects	379316	405	2544	3	2.7	1.10	0.66	1.27	0.36	3.13	2188	0.95
TAR_dup	aneurism	379893	407	1967	1	2.1	0.48	0.67	0.72	0.08	2.57	2207	0.95
TAR_dup	heartfailure	376779	402	5081	6	5.4	1.11	0.68	1.18	0.49	2.38	2247	0.95
TAR_dup	osteoporosis	370255	396	11605	12	12.4	0.97	0.69	1.12	0.60	1.91	2278	0.95
TAR_dup	inflammatory_bowel_disease	363249	387	18611	21	19.8	1.06	0.70	1.09	0.68	1.64	2300	0.96
TAR_dup	stroke_derived2017	373032	398	8828	10	9.4	1.06	0.73	1.11	0.56	1.96	2392	0.96
TAR_dup	ureter_bladder	333280	355	48580	53	51.7	1.02	0.76	1.05	0.77	1.38	2482	0.97
TAR_dup	renal	374563	400	7297	8	7.8	1.03	0.81	1.09	0.51	2.01	2605	0.97
TAR_dup	migraine	368169	395	13691	13	14.7	0.89	0.84	0.95	0.52	1.56	2702	0.97
TAR_dup	ischaemic_heart_disease_not_MI	365521	392	16339	16	17.5	0.92	0.85	0.95	0.56	1.52	2735	0.97
TAR_dup	died_to_2016	370880	397	10980	11	11.8	0.94	0.86	0.95	0.50	1.63	2768	0.97
TAR_dup	heart_valve	373476	400	8384	8	9.0	0.89	0.91	0.96	0.45	1.78	2903	0.98
TAR_dup	nasal	378665	405	3195	3	3.4	0.88	0.94	1.04	0.29	2.56	2995	0.99
TAR_dup	cerebrovascular_disease_not_stroke	377526	404	4334	4	4.6	0.86	0.99	1.01	0.34	2.27	3096	1.00
TAR_dup	respiratory	361088	386	20772	22	22.2	0.99	0.99	1.00	0.63	1.50	3113	1.00
TAR_dup	connective_tissue_disorder	372499	399	9361	9	10.0	0.90	0.99	1.00	0.49	1.79	3117	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
WBS_dup	glaucoma	374936	12	6529	1	0.2	4.49	0.069	7.68	0.81	34.20	399	0.54
WBS_dup	cataract	359613	11	21852	2	0.7	2.69	0.079	4.45	0.82	16.99	445	0.55
WBS_dup	anaemia	362323	11	19142	2	0.6	3.07	0.098	3.87	0.74	13.44	522	0.59
WBS_dup	gout	374374	12	7091	1	0.2	4.14	0.10	5.99	0.62	28.14	537	0.61
WBS_dup	arrhythmia	357305	11	24160	2	0.7	2.43	0.12	3.55	0.67	12.73	607	0.63
WBS_dup	stroke_derived2017	372646	12	8819	1	0.3	3.33	0.14	4.88	0.52	21.11	655	0.65
WBS_dup	connective_tissue_disorder	372112	12	9353	1	0.3	3.14	0.14	4.71	0.50	20.38	674	0.67
WBS_dup	gastrointestinal_ulcer	371112	12	10353	1	0.3	2.83	0.16	4.26	0.46	17.97	732	0.70
WBS_dup	varicose_veins	369612	12	11853	1	0.4	2.48	0.20	3.74	0.40	15.77	839	0.74
WBS_dup	asthma	332141	13	49324	0	1.9	0.00	0.21	0.24	0.00	1.79	865	0.75
WBS_dup	ureter_bladder	332938	13	48527	0	1.9	0.00	0.23	0.25	0.00	1.92	937	0.78
WBS_dup	digestive	299620	12	81845	1	3.3	0.36	0.27	0.42	0.05	1.77	1058	0.80
WBS_dup	any_Ca	331769	13	49696	0	1.9	0.00	0.28	0.28	0.00	2.16	1089	0.80
WBS_dup	aneurism	379499	13	1966	0	0.1	0.00	0.29	7.57	0.06	62.46	1121	0.80
WBS_dup	ischaemic_heart_disease_not_MI	365141	12	16324	1	0.5	1.80	0.31	2.78	0.29	12.35	1183	0.81
WBS_dup	prostate_hyperplasia	367017	12	14448	1	0.5	2.03	0.33	2.85	0.27	16.95	1248	0.82
WBS_dup	paralytic_syndromes	379505	13	1960	0	0.1	0.00	0.34	5.85	0.05	44.66	1275	0.83
WBS_dup	endocrine_other	379105	13	2360	0	0.1	0.00	0.34	5.86	0.05	45.18	1277	0.83
WBS_dup	neuropathies	365561	12	15904	1	0.5	1.85	0.34	2.56	0.28	10.83	1281	0.82
WBS_dup	low_WBC_count	378500	13	2965	0	0.1	0.00	0.37	5.05	0.04	38.12	1368	0.85
WBS_dup	coagulation_defects	378924	13	2541	0	0.1	0.00	0.38	4.84	0.04	36.78	1407	0.85
WBS_dup	nasal	378273	13	3192	0	0.1	0.00	0.39	4.74	0.04	36.25	1422	0.85
WBS_dup	diabetes_insulin_dependent	378871	13	2594	0	0.1	0.00	0.40	4.51	0.04	34.45	1467	0.85
WBS_dup	inflammatory_bowel_disease	362874	12	18591	1	0.6	1.58	0.41	2.19	0.24	9.18	1500	0.86
WBS_dup	gastric_reflux	347170	13	34295	0	1.3	0.00	0.42	0.37	0.00	2.81	1513	0.87
WBS_dup	osteoarthritis	312759	12	68706	1	2.6	0.43	0.47	0.55	0.06	2.42	1678	0.88
WBS_dup	cerebrovascular_disease_not_stroke	377135	13	4330	0	0.1	0.00	0.48	3.40	0.03	26.91	1697	0.88
WBS_dup	heartfailure	376390	13	5075	0	0.2	0.00	0.56	2.68	0.02	21.28	1903	0.92
WBS_dup	congenital	376438	13	5027	0	0.2	0.00	0.57	2.57	0.02	19.47	1935	0.92
WBS_dup	thyroid	354325	13	27140	0	1.0	0.00	0.61	0.52	0.00	4.11	2054	0.94
WBS_dup	atherosclerotic_vascular_disease	375716	13	5749	0	0.2	0.00	0.62	2.24	0.02	17.22	2064	0.94
WBS_dup	psoriasis	375445	13	6020	0	0.2	0.00	0.63	2.17	0.02	16.42	2096	0.94
WBS_dup	diverticular_disease_intestine	354332	13	27133	0	1.0	0.00	0.63	0.53	0.00	4.15	2100	0.94
WBS_dup	diabetes_other	360731	13	20734	0	0.7	0.00	0.68	0.58	0.00	4.49	2239	0.95
WBS_dup	renal	374176	13	7289	0	0.3	0.00	0.70	1.83	0.01	13.90	2294	0.96
WBS_dup	respiratory	360715	13	20750	0	0.7	0.00	0.70	0.60	0.00	4.57	2296	0.96
WBS_dup	ovarian_cysts	374077	13	7388	0	0.3	0.00	0.72	1.76	0.01	14.95	2358	0.96
WBS_dup	heart_valve	373089	13	8376	0	0.3	0.00	0.73	1.71	0.01	13.17	2384	0.96
WBS_dup	ocular	359805	13	21660	0	0.8	0.00	0.75	0.65	0.01	4.97	2422	0.97
WBS_dup	biliary	360691	13	20774	0	0.7	0.00	0.75	0.65	0.01	5.06	2444	0.96
WBS_dup	renal_failure	373545	13	7920	0	0.3	0.00	0.76	1.61	0.01	12.64	2460	0.97
WBS_dup	allergy	322803	11	58662	2	2.0	1.00	0.78	1.23	0.24	4.17	2513	0.97
WBS_dup	hernia	333576	12	47889	1	1.7	0.61	0.81	0.81	0.09	3.48	2607	0.97
WBS_dup	osteoporosis	369872	13	11593	0	0.4	0.00	0.83	1.40	0.01	11.61	2657	0.97
WBS_dup	hepatic	372712	13	8753	0	0.3	0.00	0.83	1.39	0.01	10.59	2663	0.97
WBS_dup	sciatica	338353	12	43112	1	1.5	0.68	0.88	0.88	0.10	3.68	2820	0.98
WBS_dup	COPD	367237	13	14228	0	0.5	0.00	0.89	0.83	0.01	6.54	2862	0.98
WBS_dup	died_to_2016	370496	13	10969	0	0.4	0.00	0.92	1.16	0.01	8.89	2925	0.98
WBS_dup	heart_other	369644	13	11821	0	0.4	0.00	0.92	1.16	0.01	9.06	2931	0.98
WBS_dup	hypertension	260830	9	120635	4	4.2	0.97	0.94	1.05	0.29	3.30	2988	0.99
WBS_dup	MI_derived2017	367605	13	13860	0	0.5	0.00	0.95	0.92	0.01	7.51	3021	0.99
WBS_dup	obesity	371628	13	9837	0	0.3	0.00	0.95	1.09	0.01	8.39	3023	0.99
WBS_dup	ear	367246	13	14219	0	0.5	0.00	0.96	0.93	0.01	7.06	3030	0.99
WBS_dup	uterine_problem	339176	12	42289	1	1.5	0.69	0.96	0.96	0.10	4.80	3043	0.99
WBS_dup	venous_thromboembolic_disease	367493	13	13972	0	0.5	0.00	0.97	0.95	0.01	7.25	3068	0.99
WBS_dup	high_cholesterol	314883	11	66582	2	2.3	0.88	0.98	1.02	0.19	3.77	3078	0.99
WBS_dup	migraine	367787	13	13678	0	0.5	0.00	0.99	1.01	0.01	7.87	3116	1.00
WBS_dup	irritable_bowel_syndrome	368462	13	13003	0	0.5	0.00	1.00	1.00	0.01	7.70	3127	1.00



CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
TAR_dup	allergy	323119	327	58741	81	59.4	1.29	0.0092	1.40	1.09	1.77	118	0.25
15q13.3dup(CHRNA7)	allergy	325161	2369	59134	474	430.8	1.08	0.047	1.11	1.00	1.22	320	0.46
16p11.2_del	allergy	322874	82	58681	21	14.9	1.33	0.081	1.56	0.94	2.46	459	0.55
15q24_dup	allergy	322797	5	58663	3	0.9	2.44	0.094	3.43	0.79	13.02	504	0.59
3q29_del	allergy	322798	6	58663	3	1.1	2.17	0.10	3.28	0.77	11.68	528	0.60
15q11.2_dup	allergy	324440	1648	58929	269	299.3	0.91	0.12	0.90	0.79	1.03	588	0.63
2q13del(NPHP1)	allergy	324789	1997	58993	333	362.7	0.93	0.12	0.91	0.81	1.02	612	0.63
2q21.1_dup	allergy	322835	43	58672	12	7.8	1.42	0.13	1.66	0.85	3.04	649	0.64
15q13.3del(CHRNA7)	allergy	322799	7	58663	3	1.3	1.95	0.13	2.86	0.69	9.70	651	0.65
7q11.23_dup_distal	allergy	322814	22	58661	1	4.0	0.28	0.15	0.35	0.04	1.37	693	0.68
1q21.1_dup	allergy	322928	136	58692	32	24.7	1.24	0.17	1.32	0.88	1.91	756	0.72
15q13.3_del	allergy	322821	29	58668	8	5.3	1.41	0.20	1.69	0.74	3.49	841	0.74
2q11.2_dup	allergy	322812	20	58666	6	3.6	1.50	0.24	1.75	0.66	4.02	965	0.78
17p12_CMT1A_dup	allergy	322886	94	58682	22	17.1	1.23	0.27	1.31	0.80	2.03	1049	0.80
2q13dup(NPHP1)	allergy	324353	1561	58967	307	283.8	1.07	0.27	1.07	0.95	1.21	1051	0.80
10q23_dup	allergy	322797	5	58662	2	0.9	1.86	0.27	2.46	0.44	10.24	1061	0.80
15q11q13dup_BP3_BP5	allergy	322801	9	58660	0	1.6	0.00	0.27	0.27	0.00	2.16	1069	0.80
15q13.3_dup	allergy	322989	197	58687	27	35.8	0.78	0.28	0.81	0.53	1.18	1079	0.80
16p12.1_dup	allergy	322950	158	58694	34	28.7	1.15	0.35	1.20	0.81	1.71	1321	0.83
13q12.12_dup	allergy	322976	184	58688	28	33.4	0.86	0.37	0.84	0.55	1.22	1376	0.85
22q11.2_dup	allergy	323013	221	58705	45	40.2	1.10	0.38	1.16	0.83	1.57	1420	0.85
15q11.2_del	allergy	324109	1317	58913	253	239.4	1.05	0.39	1.06	0.93	1.21	1440	0.85
17q11.2_del_NF1	allergy	322799	7	58662	2	1.3	1.45	0.39	1.95	0.36	7.33	1444	0.85
16p13.11_dup	allergy	323463	671	58772	112	121.9	0.93	0.43	0.92	0.75	1.12	1540	0.87
2q13_dup	allergy	322850	58	58673	13	10.5	1.19	0.44	1.27	0.67	2.23	1572	0.87
15q11q13del_BP3-BP4	allergy	322803	11	58663	3	2.0	1.39	0.44	1.64	0.42	4.98	1575	0.87
10q11.21q11.23_dup	allergy	322828	36	58664	4	6.5	0.65	0.44	0.69	0.22	1.66	1578	0.87
16p11.2_dup	allergy	322907	115	58676	16	20.9	0.79	0.46	0.83	0.48	1.35	1651	0.88
NRXN1_del	allergy	322928	136	58681	21	24.7	0.87	0.47	0.85	0.52	1.31	1672	0.88
17q12_del	allergy	322799	7	58660	0	1.3	0.00	0.47	0.40	0.00	3.29	1673	0.88
22q11.2_del	allergy	322800	8	58662	2	1.5	1.30	0.50	1.67	0.31	6.09	1765	0.89
Potocki_Lupski	allergy	322796	4	58661	1	0.7	1.30	0.50	1.97	0.20	10.79	1767	0.89
17q12_dup	allergy	322879	87	58672	12	15.8	0.79	0.51	0.82	0.43	1.43	1783	0.89
22q11.2distal_del	allergy	322796	4	58661	1	0.7	1.30	0.55	1.82	0.18	9.87	1882	0.91
1q21.1_del	allergy	322884	92	58674	14	16.7	0.86	0.55	0.85	0.47	1.43	1886	0.91
17p12_HNPP_del	allergy	322981	189	58690	30	34.3	0.89	0.55	0.89	0.60	1.29	1889	0.91
2q13_del	allergy	322834	42	58669	9	7.6	1.15	0.56	1.24	0.58	2.41	1904	0.91
TAR_del	allergy	322849	57	58672	12	10.4	1.13	0.58	1.19	0.62	2.12	1967	0.93
3q29_dup	allergy	322797	5	58660	0	0.91	0.00	0.61	0.51	0.00	4.52	2053	0.94
8p23.1_dup	allergy	322797	5	58660	0	0.9	0.00	0.63	0.52	0.00	4.67	2102	0.94
13q12.12_del	allergy	322865	73	58671	11	13.3	0.85	0.65	0.87	0.44	1.55	2146	0.95
16p13.11_del	allergy	322895	103	58681	21	18.7	1.10	0.68	1.10	0.68	1.72	2233	0.95
22q11.2distal_dup	allergy	322803	11	58661	1	2.0	0.54	0.72	0.74	0.08	3.11	2327	0.96
2q21.1_del	allergy	322827	35	58665	5	6.4	0.81	0.72	0.85	0.31	1.93	2343	0.96
16p11.2distal_del	allergy	322839	47	58667	7	8.5	0.84	0.76	0.89	0.38	1.81	2475	0.96
WBS_dup	allergy	322803	11	58662	2	2.0	1.00	0.78	1.23	0.24	4.17	2513	0.97
10q11.21q11.23_del	allergy	322839	47	58669	9	8.5	1.05	0.78	1.10	0.52	2.12	2533	0.97
13q12del_CRYL1	allergy	323100	308	58715	55	56.0	0.99	0.82	0.97	0.72	1.28	2645	0.97
PWS_dup	allergy	322806	14	58662	2	2.5	0.81	0.88	0.90	0.18	2.95	2819	0.98
13q12dup_CRYL1	allergy	322801	9	58661	1	1.6	0.65	0.90	0.90	0.10	3.92	2884	0.98
16p11.2distal_dup	allergy	322899	107	58680	20	19.4	1.02	0.93	0.98	0.58	1.55	2946	0.98
2q11.2_del	allergy	322817	25	58664	4	4.5	0.90	0.94	1.04	0.33	2.58	2975	0.99
16p12.1_del	allergy	322992	200	58695	35	36.3	0.97	0.97	0.99	0.68	1.40	3051	0.99
15q11q13dup_BP3_BP4	allergy	322835	43	58667	7	7.8	0.91	0.99	0.99	0.42	2.04	3107	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2_del	anaemia	362396	84	19159	19	4.4	3.68	2.15E-06	4.03	2.38	6.48	9	0.00075
17p12_CMT1A_dup	anaemia	362411	99	19157	17	5.2	2.92	6.70E-05	3.30	1.91	5.36	18	0.012
16p13.11_del	anaemia	362423	111	19153	13	5.9	2.09	0.014	2.21	1.19	3.75	146	0.29
16p12.1_del	anaemia	362527	215	19160	20	11.4	1.70	0.027	1.74	1.07	2.68	226	0.37
17q11.2_del_NF1	anaemia	362319	7	19142	2	0.4	4.43	0.035	6.28	1.17	23.69	266	0.41
1q21.1_dup	anaemia	362465	153	19155	15	8.1	1.78	0.040	1.82	1.03	2.97	288	0.43
3q29_dup	anaemia	362316	4	19141	1	0.21	3.99	0.067	8.54	0.83	48.52	386	0.54
10q11.21q11.23_del	anaemia	362362	50	19146	6	2.6	2.14	0.089	2.20	0.87	4.65	483	0.58
WBS_dup	anaemia	362323	11	19142	2	0.6	3.07	0.098	3.87	0.74	13.44	522	0.59
Potocki Lupski	anaemia	362316	4	19141	1	0.2	3.99	0.12	5.72	0.57	31.09	586	0.63
PWS_dup	anaemia	362326	14	19142	2	0.7	2.49	0.13	3.34	0.66	11.03	629	0.64
10q23_dup	anaemia	362318	6	19141	1	0.3	2.85	0.14	4.97	0.51	24.37	666	0.66
15q13.3_dup	anaemia	362520	208	19156	16	11.0	1.42	0.14	1.49	0.87	2.39	671	0.66
16p11.2_dup	anaemia	362433	121	19150	10	6.4	1.52	0.18	1.58	0.79	2.83	792	0.73
2q13del(NPHP1)	anaemia	364539	2227	19243	103	117.6	0.88	0.19	0.88	0.72	1.06	800	0.73
17p12_HNPP_del	anaemia	362516	204	19155	15	10.8	1.37	0.19	1.44	0.82	2.34	808	0.74
3q29_del	anaemia	362320	8	19141	1	0.4	2.21	0.25	3.28	0.35	14.76	983	0.78
17q12_dup	anaemia	362404	92	19147	7	4.9	1.41	0.25	1.60	0.69	3.16	987	0.78
2q13_del	anaemia	362359	47	19144	4	2.5	1.56	0.26	1.84	0.60	4.36	1023	0.79
22q11.2_del	anaemia	362321	9	19141	1	0.5	1.99	0.28	2.98	0.32	13.10	1087	0.80
16p13.11_dup	anaemia	363062	750	19173	33	39.6	0.84	0.30	0.84	0.58	1.16	1145	0.81
15q11.2_del	anaemia	363795	1483	19227	87	78.4	1.10	0.31	1.12	0.90	1.38	1199	0.81
2q11.2_dup	anaemia	362336	24	19142	2	1.3	1.53	0.35	1.98	0.40	6.14	1310	0.83
16p11.2distal_del	anaemia	362362	50	19144	4	2.6	1.48	0.36	1.63	0.53	3.85	1331	0.84
2q21.1_del	anaemia	362349	37	19143	3	2.0	1.49	0.38	1.70	0.46	4.47	1398	0.85
15q13.3_del	anaemia	362346	34	19143	3	1.8	1.62	0.40	1.66	0.45	4.39	1475	0.85
13q12del_CRYL1	anaemia	362655	343	19160	20	18.1	1.10	0.44	1.20	0.74	1.82	1579	0.87
2q11.2_del	anaemia	362339	27	19142	2	1.4	1.37	0.45	1.72	0.35	5.26	1606	0.87
22q11.2distal_dup	anaemia	362323	11	19141	1	0.6	1.66	0.46	2.03	0.22	8.73	1642	0.88
15q11q13dup_BP3_BP4	anaemia	362361	49	19141	1	2.6	0.40	0.50	0.60	0.07	2.22	1769	0.89
TAR_dup	anaemia	362703	391	19157	17	20.7	0.83	0.54	0.86	0.51	1.35	1862	0.91
TAR_del	anaemia	362377	65	19144	4	3.4	1.16	0.55	1.36	0.44	3.16	1887	0.91
2q13_dup	anaemia	362381	69	19142	2	3.6	0.56	0.56	0.70	0.15	2.03	1921	0.92
13q12.12_del	anaemia	362391	79	19145	5	4.2	1.19	0.59	1.28	0.47	2.78	1986	0.93
15q11.2_dup	anaemia	364126	1814	19243	103	95.9	1.07	0.61	1.05	0.86	1.28	2041	0.94
NRXN1_del	anaemia	362463	151	19146	6	8.0	0.76	0.62	0.82	0.34	1.67	2062	0.94
2q21.1_dup	anaemia	362364	52	19143	3	2.7	1.09	0.64	1.30	0.36	3.36	2136	0.94
16p11.2distal_dup	anaemia	362432	120	19147	7	6.3	1.10	0.69	1.17	0.51	2.29	2256	0.95
1q21.1_del	anaemia	362412	100	19146	6	5.3	1.13	0.69	1.18	0.48	2.42	2268	0.95
8p23.1_dup	anaemia	362317	5	19140	0	0.3	0.00	0.72	1.76	0.01	15.71	2367	0.96
7q11.23_dup_distal	anaemia	362334	22	19141	1	1.2	0.87	0.75	1.33	0.15	5.16	2425	0.97
2q13dup(NPHP1)	anaemia	364089	1777	19231	91	93.9	0.97	0.76	0.97	0.78	1.19	2478	0.96
15q11q13del_BP3-BP4	anaemia	362326	14	19140	0	0.7	0.00	0.78	0.69	0.01	5.19	2534	0.97
15q13.3dup(CHRNA7)	anaemia	365011	2699	19284	144	142.6	1.01	0.80	1.02	0.86	1.21	2577	0.97
10q11.21q11.23_dup	anaemia	362351	39	19141	1	2.1	0.50	0.80	0.82	0.09	3.06	2593	0.97
13q12.12_dup	anaemia	362513	201	19151	11	10.6	1.03	0.82	1.07	0.56	1.86	2638	0.97
22q11.2distal_del	anaemia	362317	5	19140	0	0.3	0.00	0.83	1.41	0.01	12.98	2656	0.97
16p12.1_dup	anaemia	362494	182	19150	10	9.6	1.04	0.84	1.07	0.54	1.90	2687	0.97
17q12_del	anaemia	362319	7	19140	0	0.4	0.00	0.88	1.25	0.01	10.31	2817	0.98
13q12dup_CRYL1	anaemia	362322	10	19140	0	0.5	0.00	0.89	0.82	0.01	6.40	2832	0.98
15q13.3del(CHRNA7)	anaemia	362322	10	19140	0	0.5	0.00	0.93	0.89	0.01	6.97	2960	0.99
22q11.2_dup	anaemia	362565	253	19153	13	13.4	0.97	0.94	0.98	0.54	1.63	2965	0.99
15q24_dup	anaemia	362320	8	19140	0	0.4	0.00	0.95	1.09	0.01	8.82	3025	0.99
15q11q13dup_BP3_BP5	anaemia	362321	9	19140	0	0.5	0.00	0.96	0.93	0.01	7.41	3032	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
NRXN1_del	aneurism	379638	152	1971	5	0.8	6.18	0.00042	7.59	2.82	16.42	32	0.041
22q11.2distal_del	aneurism	379490	4	1967	1	0.0	38.80	0.0013	104.85	9.71	673.61	46	0.092
2q21.1_del	aneurism	379524	38	1968	2	0.2	9.70	0.0069	11.20	2.26	34.54	94	0.23
2q13_del	aneurism	379536	50	1967	1	0.3	3.80	0.088	6.22	0.69	23.71	482	0.57
16p11.2_dup	aneurism	379615	129	1968	2	0.7	2.96	0.094	3.66	0.76	10.57	503	0.59
15q24_dup	aneurism	379494	8	1966	0	0.0	0.00	0.11	33.02	0.25	274.22	571	0.63
3q29_dup	aneurism	379491	5	1966	0	0.03	0.00	0.12	33.31	0.25	310.45	575	0.63
10q11.21q11.23_del	aneurism	379541	55	1967	1	0.3	3.46	0.12	4.99	0.56	19.05	615	0.63
TAR_del	aneurism	379554	68	1967	1	0.4	2.81	0.16	4.24	0.48	15.91	716	0.69
Potocki_Lupski	aneurism	379491	5	1966	0	0.0	0.00	0.18	16.20	0.12	174.61	789	0.73
15q11q13dup_BP3_BP5	aneurism	379495	9	1966	0	0.0	0.00	0.19	14.74	0.11	126.55	807	0.74
8p23.1_dup	aneurism	379491	5	1966	0	0.0	0.00	0.22	12.25	0.09	119.98	892	0.76
10q23_dup	aneurism	379493	7	1966	0	0.0	0.00	0.22	11.88	0.09	115.77	902	0.76
13q12dup_CRYL1	aneurism	379496	10	1966	0	0.1	0.00	0.23	10.76	0.08	93.40	936	0.78
3q29_del	aneurism	379495	9	1966	0	0.0	0.00	0.24	10.44	0.08	90.55	948	0.78
16p13.11_dup	aneurism	380263	777	1972	6	4.0	1.49	0.24	1.66	0.68	3.31	949	0.78
17q12_del	aneurism	379493	7	1966	0	0.0	0.00	0.24	10.43	0.08	94.86	952	0.78
15q11q13del_BP3-BP4	aneurism	379500	14	1966	0	0.1	0.00	0.24	9.89	0.08	76.78	968	0.78
22q11.2_del	aneurism	379496	10	1966	0	0.1	0.00	0.25	9.66	0.07	84.26	999	0.78
PWS_dup	aneurism	379502	16	1966	0	0.1	0.00	0.25	9.47	0.07	79.51	1005	0.78
1q21.1_del	aneurism	379591	105	1967	1	0.5	1.83	0.26	3.01	0.34	11.09	1021	0.79
15q13.3del(CHRNA7)	aneurism	379496	10	1966	0	0.1	0.00	0.26	8.75	0.07	72.00	1035	0.79
WBS_dup	aneurism	379499	13	1966	0	0.1	0.00	0.29	7.57	0.06	62.46	1121	0.80
17q11.2_del_NF1	aneurism	379495	9	1966	0	0.0	0.00	0.30	7.10	0.05	60.66	1163	0.81
16p11.2distal_dup	aneurism	379612	126	1967	1	0.7	1.53	0.31	2.63	0.30	9.61	1197	0.81
22q11.2distal_dup	aneurism	379498	12	1966	0	0.1	0.00	0.32	6.26	0.05	49.86	1237	0.82
17p12_CMT1A_dup	aneurism	379601	115	1967	1	0.6	1.67	0.35	2.43	0.28	8.94	1308	0.83
22q11.2_dup	aneurism	379750	264	1968	2	1.4	1.46	0.36	1.90	0.40	5.42	1343	0.84
2q11.2_dup	aneurism	379512	26	1966	0	0.1	0.00	0.40	4.45	0.03	32.83	1474	0.85
7q11.23_dup_distal	aneurism	379509	23	1966	0	0.1	0.00	0.42	4.09	0.03	31.53	1534	0.87
2q11.2_del	aneurism	379515	29	1966	0	0.2	0.00	0.48	3.39	0.03	25.09	1691	0.88
10q11.21q11.23_dup	aneurism	379526	40	1966	0	0.2	0.00	0.49	3.20	0.03	23.03	1740	0.89
1q21.1_dup	aneurism	379653	167	1967	1	0.9	1.15	0.52	1.79	0.20	6.49	1810	0.90
13q12.12_dup	aneurism	379698	212	1966	0	1.1	0.00	0.56	0.48	0.00	3.29	1908	0.91
15q13.3dup(CHRNA7)	aneurism	382313	2827	1982	16	14.7	1.09	0.57	1.16	0.68	1.82	1939	0.92
2q13dup(NPHP1)	aneurism	381343	1857	1977	11	9.6	1.14	0.59	1.18	0.62	2.01	1973	0.93
16p12.1_dup	aneurism	379678	192	1966	0	1.0	0.00	0.61	0.52	0.00	3.58	2029	0.94
15q13.3_del	aneurism	379523	37	1966	0	0.2	0.00	0.61	2.29	0.02	16.54	2035	0.94
15q11q13dup_BP3_BP4	aneurism	379536	50	1966	0	0.3	0.00	0.66	2.02	0.02	14.35	2176	0.95
TAR_dup	aneurism	379893	407	1967	1	2.1	0.48	0.67	0.72	0.08	2.57	2207	0.95
15q11.2_dup	aneurism	381395	1909	1974	8	9.9	0.81	0.69	0.87	0.41	1.61	2270	0.95
15q11.2_del	aneurism	381047	1561	1975	9	8.1	1.11	0.71	1.13	0.55	2.02	2313	0.96
16p11.2distal_del	aneurism	379540	54	1966	0	0.3	0.00	0.71	1.77	0.01	12.50	2321	0.96
2q21.1_dup	aneurism	379541	55	1966	0	0.3	0.00	0.76	1.59	0.01	11.21	2479	0.96
17p12_HNPP_del	aneurism	379704	218	1967	1	1.1	0.89	0.78	1.27	0.15	4.59	2520	0.97
16p12.1_del	aneurism	379720	234	1967	1	1.2	0.83	0.79	1.25	0.14	4.52	2563	0.97
2q13_dup	aneurism	379557	71	1966	0	0.4	0.00	0.87	1.27	0.01	8.89	2794	0.98
15q13.3_dup	aneurism	379709	223	1967	1	1.2	0.87	0.87	1.14	0.13	4.12	2799	0.98
13q12del_CRYL1	aneurism	379848	362	1967	1	1.9	0.53	0.88	0.89	0.10	3.19	2824	0.98
17q12_dup	aneurism	379585	99	1966	0	0.5	0.00	0.89	0.82	0.01	5.69	2830	0.98
2q13del(NPHP1)	aneurism	381804	2318	1978	12	12.0	1.00	0.90	1.04	0.56	1.73	2880	0.98
16p13.11_del	aneurism	379610	124	1966	0	0.6	0.00	0.92	0.87	0.01	6.02	2929	0.98
16p11.2_del	aneurism	379589	103	1966	0	0.5	0.00	0.96	0.94	0.01	6.52	3048	0.99
13q12.12_del	aneurism	379570	84	1966	0	0.4	0.00	0.98	1.03	0.01	7.17	3093	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
3q29_dup	any_Ca	331757	1	49700	4	0.15	6.14	6.23E-05	37.54	6.51	389.10	17	0.011*
16p12.1_dup	any_Ca	331933	177	49711	15	26.5	0.60	0.038	0.60	0.34	0.97	280	0.43
17p12_HNPP_del	any_Ca	331955	199	49716	20	29.8	0.70	0.053	0.65	0.40	1.00	340	0.49
16p11.2distal_dup	any_Ca	331862	106	49717	21	15.9	1.27	0.071	1.58	0.96	2.48	408	0.54
15q13.3_del	any_Ca	331792	36	49697	1	5.4	0.21	0.075	0.29	0.03	1.11	430	0.55
13q12.12_del	any_Ca	331823	67	49713	17	10.0	1.55	0.079	1.66	0.94	2.77	450	0.55
15q11q13dup_BP3_BP4	any_Ca	331804	48	49698	2	7.2	0.31	0.081	0.37	0.08	1.11	457	0.56
16p11.2_dup	any_Ca	331877	121	49706	10	18.1	0.59	0.086	0.59	0.30	1.07	475	0.57
8p23.1_dup	any_Ca	331759	3	49698	2	0.4	3.07	0.11	4.19	0.68	22.07	565	0.63
TAR_dup	any_Ca	332123	367	49737	41	55.0	0.77	0.13	0.78	0.56	1.07	619	0.63
2q21.1_del	any_Ca	331794	38	49698	2	5.7	0.38	0.14	0.42	0.09	1.27	665	0.66
2q13dup(NPHP1)	any_Ca	333400	1644	49920	224	246.2	0.92	0.21	0.91	0.79	1.05	875	0.75
2q11.2_dup	any_Ca	331777	21	49701	5	3.1	1.48	0.23	1.85	0.64	4.52	943	0.78
22q11.2distal_del	any_Ca	331760	4	49697	1	0.6	1.54	0.25	3.48	0.34	19.66	988	0.78
16p11.2_del	any_Ca	331845	89	49710	14	13.3	1.04	0.26	1.41	0.77	2.40	1019	0.79
16p13.11_dup	any_Ca	332430	674	49805	109	101.0	1.07	0.26	1.13	0.91	1.38	1029	0.79
17p12_CMT1A_dup	any_Ca	331854	98	49714	18	14.7	1.19	0.27	1.35	0.79	2.18	1045	0.80
2q13del(NPHP1)	any_Ca	333800	2044	49982	286	306.1	0.94	0.27	0.93	0.82	1.06	1066	0.80
WBS_dup	any_Ca	331769	13	49696	0	1.9	0.00	0.28	0.28	0.00	2.16	1089	0.80
PWS_dup	any_Ca	331772	16	49696	0	2.4	0.00	0.30	0.29	0.00	2.26	1161	0.81
TAR_del	any_Ca	331814	58	49707	11	8.7	1.22	0.31	1.41	0.70	2.59	1207	0.81
13q12.12_dup	any_Ca	331937	181	49727	31	27.1	1.12	0.32	1.22	0.82	1.77	1218	0.82
1q21.1_dup	any_Ca	331900	144	49720	24	21.6	1.10	0.39	1.22	0.77	1.84	1430	0.85
13q12dup_CRYL1	any_Ca	331764	8	49698	2	1.2	1.54	0.39	1.96	0.36	7.33	1434	0.85
13q12del_CRYL1	any_Ca	332070	314	49745	49	47.0	1.04	0.40	1.14	0.83	1.53	1473	0.85
Potocki_Lupski	any_Ca	331760	4	49697	1	0.6	1.54	0.44	2.21	0.22	12.40	1580	0.87
15q13.3del(CHRNA7)	any_Ca	331766	10	49696	0	1.5	0.00	0.45	0.39	0.00	3.07	1607	0.87
17q11.2_del_NF1	any_Ca	331763	7	49698	2	1.0	1.71	0.46	1.78	0.33	6.79	1640	0.88
16p12.1_del	any_Ca	331958	202	49729	33	30.3	1.08	0.47	1.15	0.78	1.64	1683	0.88
1q21.1_del	any_Ca	331852	96	49706	10	14.4	0.72	0.48	0.80	0.39	1.45	1715	0.88
2q21.1_dup	any_Ca	331806	50	49701	5	7.5	0.70	0.53	0.76	0.28	1.69	1834	0.90
16p13.11_del	any_Ca	331867	111	49709	13	16.6	0.80	0.54	0.84	0.45	1.44	1865	0.91
17q12_del	any_Ca	331762	6	49697	1	0.9	1.10	0.56	1.76	0.18	8.68	1925	0.91
15q11.2_dup	any_Ca	333420	1664	49949	253	249.3	1.01	0.57	1.04	0.91	1.19	1936	0.92
22q11.2_del	any_Ca	331766	10	49696	0	1.5	0.00	0.58	0.49	0.00	3.91	1969	0.93
2q13_del	any_Ca	331803	47	49700	4	7.0	0.60	0.59	0.77	0.25	1.84	1975	0.93
2q13_dup	any_Ca	331820	64	49703	7	9.6	0.76	0.59	0.81	0.35	1.64	1987	0.93
2q11.2_del	any_Ca	331783	27	49698	2	4.0	0.53	0.59	0.71	0.14	2.19	1992	0.93
22q11.2distal_dup	any_Ca	331766	10	49698	2	1.5	1.28	0.61	1.45	0.28	5.08	2042	0.94
22q11.2_dup	any_Ca	331994	238	49724	28	35.6	0.81	0.64	0.91	0.60	1.33	2120	0.94
15q24_dup	any_Ca	331764	8	49696	0	1.2	0.00	0.65	0.55	0.00	4.61	2162	0.95
15q13.3_dup	any_Ca	331955	199	49721	25	29.8	0.86	0.67	0.91	0.59	1.36	2191	0.95
15q11q13dup_BP3_BP5	any_Ca	331764	8	49697	1	1.2	0.85	0.67	1.51	0.15	7.22	2198	0.95
10q23_dup	any_Ca	331763	7	49696	0	1.0	0.00	0.67	0.56	0.00	4.85	2200	0.95
10q11.21q11.23_del	any_Ca	331806	50	49702	6	7.5	0.82	0.72	0.86	0.34	1.84	2364	0.96
16p11.2distal_del	any_Ca	331805	49	49701	5	7.3	0.71	0.74	0.86	0.31	1.92	2398	0.96
7q11.23_dup_distal	any_Ca	331777	21	49698	2	3.1	0.67	0.74	0.80	0.16	2.55	2408	0.96
3q29_del	any_Ca	331764	8	49697	1	1.2	0.85	0.77	1.30	0.14	5.92	2506	0.97
15q11.2_del	any_Ca	333126	1370	49896	200	205.2	0.98	0.88	0.99	0.85	1.15	2806	0.98
15q13.3dup(CHRNA7)	any_Ca	334235	2479	50060	364	371.3	0.98	0.89	0.99	0.89	1.11	2827	0.98
15q11q13del_BP3-BP4	any_Ca	331769	13	49697	1	1.9	0.55	0.89	0.89	0.10	3.73	2853	0.98
10q11.21q11.23_dup	any_Ca	331792	36	49700	4	5.4	0.77	0.89	1.07	0.34	2.61	2857	0.98
17q12_dup	any_Ca	331843	87	49708	12	13.0	0.93	0.95	0.98	0.51	1.72	3002	0.99
NRXN1_del	any_Ca	331894	138	49715	19	20.7	0.93	0.98	1.01	0.60	1.59	3085	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p12.1_del	arrhythmia	357504	210	24183	25	14.2	1.68	0.0069	1.86	1.20	2.78	93	0.23
13q12dup_CRYL1	arrhythmia	357301	7	24161	3	0.5	4.74	0.0071	8.13	1.91	28.42	98	0.23
PWS_dup	arrhythmia	357307	13	24161	3	0.9	2.96	0.014	6.31	1.54	19.86	148	0.29
1q21.1_del	arrhythmia	357388	94	24170	12	6.4	1.79	0.022	2.16	1.13	3.80	198	0.35
17p12_CMT1A_dup	arrhythmia	357397	103	24171	13	7.0	1.77	0.030	2.00	1.07	3.45	244	0.39
22q11.2distal_dup	arrhythmia	357303	9	24161	3	0.6	3.95	0.032	4.76	1.17	15.35	254	0.39
16p11.2_del	arrhythmia	357386	92	24169	11	6.2	1.69	0.040	2.05	1.04	3.68	292	0.43
13q12del_CRYL1	arrhythmia	357627	333	24188	30	22.5	1.30	0.049	1.49	1.00	2.13	324	0.47
3q29_del	arrhythmia	357301	7	24160	2	0.5	3.51	0.056	5.24	0.95	20.48	354	0.50
16p11.2distal_dup	arrhythmia	357409	115	24170	12	7.8	1.49	0.071	1.80	0.95	3.14	406	0.54
2q13del(NPHP1)	arrhythmia	359456	2162	24326	168	146.3	1.14	0.079	1.16	0.98	1.35	446	0.55
15q11q13del_BP3-BP4	arrhythmia	357306	12	24160	2	0.8	2.26	0.099	3.81	0.74	13.00	524	0.59
WBS_dup	arrhythmia	357305	11	24160	2	0.7	2.43	0.12	3.55	0.67	12.73	607	0.63
7q11.23_dup_distal	arrhythmia	357314	20	24161	3	1.4	2.06	0.12	2.79	0.73	7.92	609	0.63
15q24_dup	arrhythmia	357301	7	24159	1	0.5	1.97	0.13	5.26	0.54	25.03	624	0.64
1q21.1_dup	arrhythmia	357447	153	24173	15	10.3	1.41	0.13	1.54	0.87	2.54	633	0.64
10q11.21q11.23_del	arrhythmia	357344	50	24164	6	3.4	1.69	0.16	1.90	0.75	4.06	730	0.70
17q12_dup	arrhythmia	357383	89	24168	10	6.0	1.59	0.16	1.63	0.80	2.99	731	0.70
2q21.1_dup	arrhythmia	357348	54	24159	1	3.7	0.29	0.18	0.39	0.04	1.44	794	0.73
NRXN1_del	arrhythmia	357438	144	24171	13	9.7	1.31	0.22	1.45	0.79	2.46	896	0.76
10q23_dup	arrhythmia	357300	6	24159	1	0.4	2.26	0.22	3.70	0.36	19.69	914	0.77
15q13.3_dup	arrhythmia	357499	205	24177	19	13.9	1.34	0.23	1.35	0.82	2.10	934	0.78
16p11.2distal_del	arrhythmia	357347	53	24159	1	3.6	0.29	0.24	0.42	0.05	1.57	947	0.78
2q13_dup	arrhythmia	357363	69	24160	2	4.7	0.44	0.27	0.52	0.11	1.53	1055	0.80
TAR_del	arrhythmia	357357	63	24164	6	4.3	1.37	0.29	1.60	0.64	3.39	1133	0.81
22q11.2_del	arrhythmia	357303	9	24159	1	0.6	1.58	0.29	2.92	0.30	13.60	1137	0.81
2q11.2_dup	arrhythmia	357320	26	24158	0	1.8	0.00	0.31	0.30	0.00	2.19	1177	0.81
16p13.11_dup	arrhythmia	358022	728	24213	55	49.2	1.11	0.31	1.16	0.87	1.51	1208	0.81
TAR_dup	arrhythmia	357672	378	24188	30	25.6	1.16	0.32	1.22	0.82	1.74	1217	0.82
2q21.1_del	arrhythmia	357330	36	24162	4	2.4	1.58	0.32	1.72	0.55	4.21	1222	0.82
15q13.3del(CHRNA7)	arrhythmia	357303	9	24159	1	0.6	1.58	0.37	2.44	0.26	10.99	1358	0.84
15q13.3dup(CHRNA7)	arrhythmia	359949	2655	24346	188	179.6	1.04	0.37	1.07	0.92	1.24	1365	0.85
16p13.11_del	arrhythmia	357413	119	24163	5	8.0	0.64	0.43	0.72	0.27	1.55	1542	0.87
15q11.2_del	arrhythmia	358770	1476	24252	94	99.8	0.95	0.47	0.93	0.74	1.14	1658	0.88
17q11.2_del_NF1	arrhythmia	357302	8	24159	1	0.5	1.75	0.47	2.02	0.21	9.30	1671	0.88
13q12.12_dup	arrhythmia	357491	197	24173	15	13.3	1.12	0.47	1.22	0.69	1.99	1681	0.88
2q13dup(NPHP1)	arrhythmia	359036	1742	24284	126	117.8	1.07	0.51	1.06	0.88	1.27	1778	0.89
17p12_HNPP_del	arrhythmia	357501	207	24170	12	14.0	0.87	0.59	0.86	0.46	1.46	1978	0.93
2q11.2_del	arrhythmia	357321	27	24160	2	1.8	1.09	0.63	1.40	0.28	4.38	2094	0.94
3q29_dup	arrhythmia	357299	5	24158	0	0.34	0.00	0.66	2.06	0.02	18.79	2177	0.95
22q11.2_dup	arrhythmia	357543	249	24175	17	16.8	1.01	0.72	1.10	0.65	1.74	2325	0.96
22q11.2distal_del	arrhythmia	357299	5	24158	0	0.3	0.00	0.72	1.77	0.01	16.74	2355	0.96
16p11.2_dup	arrhythmia	357416	122	24167	9	8.2	1.08	0.73	1.13	0.54	2.08	2386	0.96
15q11.2_dup	arrhythmia	359095	1801	24274	116	121.7	0.96	0.76	0.97	0.80	1.17	2481	0.97
2q13_del	arrhythmia	357342	48	24161	3	3.2	0.93	0.78	1.18	0.32	3.08	2516	0.97
10q11.21q11.23_dup	arrhythmia	357332	38	24160	2	2.6	0.79	0.80	1.19	0.24	3.60	2574	0.97
15q11q13dup_BP3_BP4	arrhythmia	357341	47	24161	3	3.2	0.95	0.81	1.15	0.31	3.00	2613	0.97
Potocki_Lupski	arrhythmia	357299	5	24158	0	0.3	0.00	0.87	1.28	0.01	11.99	2791	0.98
13q12.12_del	arrhythmia	357373	79	24163	5	5.3	0.94	0.90	0.95	0.35	2.07	2874	0.98
16p12.1_dup	arrhythmia	357475	181	24169	11	12.2	0.90	0.91	0.96	0.50	1.68	2892	0.98
17q12_del	arrhythmia	357301	7	24158	0	0.5	0.00	0.92	0.86	0.01	7.50	2926	0.98
15q13.3_del	arrhythmia	357329	35	24160	2	2.4	0.85	0.94	0.95	0.19	2.89	2969	0.99
8p23.1_dup	arrhythmia	357299	5	24158	0	0.3	0.00	0.97	1.06	0.01	9.77	3058	0.99
15q11q13dup_BP3_BP5	arrhythmia	357303	9	24158	0	0.6	0.00	0.97	0.94	0.01	7.81	3059	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2_del	asthma	332199	71	49356	32	10.5	2.40	1.33E-05	2.71	1.77	4.08	14	0.0030
15q13.3_del	asthma	332151	23	49338	14	3.4	2.93	0.00018	3.90	1.97	7.43	22	0.026
16p13.11_del	asthma	332226	98	49350	26	14.6	1.62	0.013	1.78	1.14	2.69	144	0.28
TAR_del	asthma	332194	66	49327	3	9.8	0.34	0.028	0.36	0.10	0.91	238	0.37
15q13.3del(CHRNA7)	asthma	332134	6	49328	4	0.9	3.09	0.031	4.21	1.16	13.95	245	0.39
10q23_dup	asthma	332132	4	49327	3	0.6	3.31	0.032	5.27	1.18	21.69	252	0.39
16p12.1_dup	asthma	332285	157	49359	35	23.3	1.41	0.037	1.50	1.03	2.14	272	0.42
10q11.21q11.23_dup	asthma	332167	39	49325	1	5.8	0.19	0.039	0.25	0.03	0.94	284	0.43
2q13_del	asthma	332167	39	49336	12	5.8	1.82	0.040	2.04	1.04	3.76	290	0.43
15q13.3_dup	asthma	332332	204	49344	20	30.3	0.69	0.047	0.65	0.40	0.99	318	0.46
3q29_del	asthma	332134	6	49327	3	0.9	2.58	0.073	3.66	0.87	12.94	421	0.55
15q11q13del_BP3-BP4	asthma	332138	10	49328	4	1.5	2.21	0.092	2.81	0.83	8.07	497	0.58
2q21.1_dup	asthma	332180	52	49327	3	7.7	0.42	0.095	0.44	0.12	1.13	507	0.59
15q11q13dup_BP3_BP5	asthma	332134	6	49327	3	0.9	2.58	0.11	3.15	0.75	11.12	552	0.62
NRXN1_del	asthma	332258	130	49351	27	19.3	1.33	0.11	1.42	0.92	2.10	556	0.63
17q12_dup	asthma	332209	81	49342	18	12.0	1.41	0.11	1.54	0.90	2.49	558	0.62
1q21.1_del	asthma	332215	87	49343	19	12.9	1.39	0.15	1.46	0.87	2.33	685	0.68
22q11.2_del	asthma	332135	7	49327	3	1.0	2.32	0.16	2.67	0.65	9.03	721	0.69
17q12_del	asthma	332133	5	49326	2	0.7	2.21	0.18	3.03	0.54	12.60	785	0.73
PWS_dup	asthma	332140	12	49328	4	1.8	1.93	0.18	2.20	0.66	6.07	786	0.73
2q13_dup	asthma	332186	58	49337	13	8.6	1.42	0.18	1.52	0.81	2.67	795	0.73
WBS_dup	asthma	332141	13	49324	0	1.9	0.00	0.21	0.24	0.00	1.79	865	0.75
22q11.2_dup	asthma	332352	224	49366	42	33.3	1.22	0.24	1.22	0.87	1.68	966	0.78
17q11.2_del_NF1	asthma	332135	7	49326	2	1.0	1.72	0.26	2.43	0.45	9.10	1038	0.79
2q11.2_dup	asthma	332149	21	49329	5	3.1	1.49	0.30	1.69	0.59	4.04	1159	0.81
16p13.11_dup	asthma	332799	671	49436	112	99.7	1.11	0.34	1.10	0.90	1.34	1293	0.83
2q21.1_del	asthma	332161	33	49331	7	4.9	1.35	0.38	1.45	0.61	3.04	1390	0.85
17p12_CMT1A_dup	asthma	332232	104	49336	12	15.4	0.80	0.39	0.78	0.41	1.34	1421	0.85
1q21.1_dup	asthma	332270	142	49350	26	21.1	1.20	0.39	1.20	0.78	1.79	1448	0.85
8p23.1_dup	asthma	332132	4	49325	1	0.6	1.55	0.39	2.41	0.24	13.06	1449	0.85
3q29_dup	asthma	332132	4	49325	1	0.59	1.55	0.41	2.32	0.23	12.57	1489	0.86
TAR_dup	asthma	332478	350	49382	58	52.0	1.10	0.42	1.12	0.84	1.47	1506	0.87
Potocki_Lupski	asthma	332132	4	49325	1	0.6	1.55	0.42	2.26	0.23	12.30	1528	0.87
15q11.2_del	asthma	333485	1357	49537	213	201.6	1.05	0.44	1.06	0.91	1.22	1567	0.87
13q12dup_CRYL1	asthma	332136	8	49326	2	1.2	1.55	0.44	1.80	0.34	6.59	1586	0.87
16p11.2_dup	asthma	332245	117	49338	14	17.4	0.83	0.50	0.83	0.46	1.39	1750	0.89
10q11.21q11.23_del	asthma	332175	47	49333	9	7.0	1.24	0.54	1.25	0.59	2.41	1853	0.91
16p11.2distal_dup	asthma	332236	108	49343	19	16.0	1.16	0.54	1.17	0.70	1.85	1858	0.91
22q11.2distal_dup	asthma	332138	10	49326	2	1.5	1.29	0.54	1.57	0.30	5.48	1867	0.91
22q11.2distal_del	asthma	332133	5	49324	0	0.7	0.00	0.58	0.48	0.00	4.29	1971	0.93
16p12.1_del	asthma	332330	202	49357	33	30.0	1.09	0.63	1.10	0.75	1.56	2095	0.94
2q13dup(NPHP1)	asthma	333748	1620	49572	248	240.6	1.03	0.68	1.03	0.90	1.17	2227	0.95
16p11.2distal_del	asthma	332176	48	49330	6	7.1	0.86	0.71	0.86	0.34	1.82	2319	0.96
7q11.23_dup_distal	asthma	332149	21	49326	2	3.1	0.67	0.72	0.79	0.16	2.46	2330	0.96
15q11q13dup_BP3_BP4	asthma	332171	43	49331	7	6.4	1.08	0.74	1.14	0.48	2.34	2407	0.96
2q11.2_del	asthma	332154	26	49327	3	3.9	0.80	0.78	0.86	0.23	2.30	2528	0.97
15q13.3dup(CHRNA7)	asthma	334607	2479	49688	364	368.1	0.99	0.79	0.99	0.88	1.10	2567	0.97
13q12del_CRYL1	asthma	332443	315	49372	48	46.8	1.02	0.83	1.03	0.75	1.38	2678	0.97
13q12.12_del	asthma	332201	73	49335	11	10.8	1.01	0.84	1.07	0.54	1.91	2690	0.97
15q24_dup	asthma	332135	7	49325	1	1.0	0.97	0.84	1.20	0.13	5.53	2706	0.97
2q13del(NPHP1)	asthma	334155	2027	49627	303	301.0	1.01	0.88	1.01	0.89	1.14	2823	0.98
17p12_HNPP_del	asthma	332320	192	49351	27	28.5	0.95	0.91	0.98	0.64	1.43	2904	0.98
13q12.12_dup	asthma	332313	185	49351	27	27.5	0.98	0.96	0.99	0.65	1.45	3035	0.99
15q11.2_dup	asthma	333795	1667	49574	250	247.6	1.01	0.96	1.00	0.87	1.14	3041	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2_del	atherosclerotic_vascular_disease	375800	97	5755	6	1.5	3.87	0.010	3.50	1.40	7.33	124	0.26
10q23_dup	atherosclerotic_vascular_disease	375709	6	5750	1	0.1	9.48	0.024	16.39	1.61	88.70	209	0.35
15q11.2_dup	atherosclerotic_vascular_disease	377579	1876	5790	41	28.8	1.42	0.035	1.42	1.03	1.91	264	0.41
15q11q13del_BP3-BP4	atherosclerotic_vascular_disease	375716	13	5750	1	0.2	4.74	0.044	9.92	1.08	41.27	309	0.45
10q11.21q11.23_dup	atherosclerotic_vascular_disease	375741	38	5751	2	0.6	3.32	0.046	5.14	1.04	15.66	314	0.45
16p12.1_del	atherosclerotic_vascular_disease	375931	228	5756	7	3.5	1.98	0.070	2.13	0.93	4.13	402	0.55
22q11.2_dup	atherosclerotic_vascular_disease	375961	258	5757	8	4.0	2.00	0.071	2.02	0.94	3.78	407	0.54
NRXN1_del	atherosclerotic_vascular_disease	375855	152	5754	5	2.3	2.11	0.073	2.44	0.91	5.23	419	0.55
2q21.1_del	atherosclerotic_vascular_disease	375741	38	5751	2	0.6	3.32	0.086	3.89	0.79	11.76	472	0.57
22q11.2distal_dup	atherosclerotic_vascular_disease	375714	11	5750	1	0.2	5.53	0.089	6.50	0.69	28.55	485	0.58
TAR_del	atherosclerotic_vascular_disease	375770	67	5751	2	1.0	1.92	0.21	2.53	0.52	7.43	882	0.75
16p13.11_del	atherosclerotic_vascular_disease	375827	124	5749	0	1.9	0.00	0.24	0.27	0.00	1.86	972	0.78
3q29_dup	atherosclerotic_vascular_disease	375708	5	5749	0	0.08	0.00	0.26	8.82	0.07	83.24	1041	0.80
17q12_dup	atherosclerotic_vascular_disease	375802	99	5749	0	1.5	0.00	0.28	0.29	0.00	2.00	1080	0.80
15q24_dup	atherosclerotic_vascular_disease	375711	8	5749	0	0.1	0.00	0.33	6.08	0.05	50.11	1260	0.82
8p23.1_dup	atherosclerotic_vascular_disease	375708	5	5749	0	0.1	0.00	0.33	6.04	0.05	54.42	1270	0.83
Potocki_Lupski	atherosclerotic_vascular_disease	375708	5	5749	0	0.1	0.00	0.35	5.55	0.04	51.87	1328	0.84
13q12.12_del	atherosclerotic_vascular_disease	375785	82	5751	2	1.3	1.58	0.37	1.89	0.39	5.51	1364	0.85
22q11.2distal_del	atherosclerotic_vascular_disease	375708	5	5749	0	0.1	0.00	0.39	4.79	0.04	48.24	1458	0.85
16p11.2_dup	atherosclerotic_vascular_disease	375831	128	5752	3	2.0	1.52	0.42	1.60	0.44	4.04	1518	0.87
15q13.3_dup	atherosclerotic_vascular_disease	375922	219	5754	5	3.4	1.48	0.45	1.42	0.53	3.02	1601	0.87
15q13.3_del	atherosclerotic_vascular_disease	375739	36	5750	1	0.6	1.79	0.46	2.00	0.22	7.65	1630	0.88
15q11q13dup_BP3_BP5	atherosclerotic_vascular_disease	375712	9	5749	0	0.1	0.00	0.46	3.60	0.03	29.69	1646	0.88
13q12.12_dup	atherosclerotic_vascular_disease	375911	208	5753	4	3.2	1.25	0.46	1.45	0.48	3.29	1650	0.88
13q12dup_CRYL1	atherosclerotic_vascular_disease	375713	10	5749	0	0.2	0.00	0.49	3.28	0.03	26.05	1727	0.89
2q13_del	atherosclerotic_vascular_disease	375753	50	5750	1	0.8	1.30	0.49	1.88	0.21	7.06	1733	0.89
3q29_del	atherosclerotic_vascular_disease	375712	9	5749	0	0.1	0.00	0.49	3.23	0.02	26.26	1744	0.89
17q12_del	atherosclerotic_vascular_disease	375710	7	5749	0	0.1	0.00	0.51	3.14	0.02	27.21	1775	0.89
2q13_dup	atherosclerotic_vascular_disease	375774	71	5749	0	1.1	0.00	0.52	0.45	0.00	3.14	1813	0.90
PWS_dup	atherosclerotic_vascular_disease	375719	16	5749	0	0.2	0.00	0.53	2.91	0.02	22.43	1831	0.90
22q11.2_del	atherosclerotic_vascular_disease	375713	10	5749	0	0.2	0.00	0.53	2.89	0.02	23.99	1847	0.90
17q11.2_del_NF1	atherosclerotic_vascular_disease	375712	9	5749	0	0.1	0.00	0.54	2.77	0.02	22.46	1873	0.91
2q13dup(NPHP1)	atherosclerotic_vascular_disease	377540	1837	5780	31	28.1	1.10	0.55	1.12	0.77	1.56	1880	0.91
2q13del(NPHP1)	atherosclerotic_vascular_disease	377995	2292	5787	38	35.1	1.08	0.56	1.10	0.79	1.50	1899	0.92
15q13.3del(CHRNA7)	atherosclerotic_vascular_disease	375713	10	5749	0	0.2	0.00	0.57	2.55	0.02	20.84	1944	0.92
17p12_HNPP_del	atherosclerotic_vascular_disease	375920	217	5751	2	3.3	0.61	0.58	0.72	0.15	2.04	1965	0.93
TAR_dup	atherosclerotic_vascular_disease	376104	401	5756	7	6.1	1.14	0.61	1.22	0.54	2.34	2026	0.94
WBS_dup	atherosclerotic_vascular_disease	375716	13	5749	0	0.2	0.00	0.62	2.24	0.02	17.22	2064	0.94
10q11.21q11.23_del	atherosclerotic_vascular_disease	375758	55	5750	1	0.8	1.18	0.63	1.53	0.17	5.76	2097	0.94
1q21.1_dup	atherosclerotic_vascular_disease	375868	165	5752	3	2.5	1.18	0.64	1.31	0.36	3.27	2113	0.94
16p11.2distal_dup	atherosclerotic_vascular_disease	375828	125	5751	2	1.9	1.04	0.65	1.36	0.28	3.90	2154	0.95
17p12_CMT1A_dup	atherosclerotic_vascular_disease	375817	114	5751	2	1.7	1.14	0.66	1.35	0.28	3.90	2165	0.95
2q21.1_dup	atherosclerotic_vascular_disease	375758	55	5749	0	0.8	0.00	0.66	0.57	0.00	3.98	2185	0.95
16p11.2distal_del	atherosclerotic_vascular_disease	375757	54	5749	0	0.8	0.00	0.67	0.58	0.00	4.03	2213	0.95
15q13.3dup(CHRNA7)	atherosclerotic_vascular_disease	378507	2804	5788	39	42.9	0.91	0.71	0.94	0.68	1.27	2312	0.96
16p12.1_dup	atherosclerotic_vascular_disease	375892	189	5752	3	2.9	1.04	0.72	1.22	0.34	3.05	2335	0.96
15q11q13dup_BP3_BP4	atherosclerotic_vascular_disease	375753	50	5749	0	0.8	0.00	0.75	0.66	0.01	4.61	2433	0.97
13q12del_CRYL1	atherosclerotic_vascular_disease	376061	358	5754	5	5.5	0.91	0.75	1.15	0.43	2.43	2434	0.97
7q11.23_dup_distal	atherosclerotic_vascular_disease	375726	23	5749	0	0.4	0.00	0.77	1.55	0.01	11.36	2510	0.97
2q11.2_dup	atherosclerotic_vascular_disease	375729	26	5749	0	0.4	0.00	0.82	1.41	0.01	10.23	2647	0.97
15q11.2_del	atherosclerotic_vascular_disease	377250	1547	5772	23	23.7	0.97	0.87	0.97	0.62	1.42	2793	0.98
16p13.11_dup	atherosclerotic_vascular_disease	376474	771	5761	12	11.8	1.02	0.91	1.03	0.56	1.73	2891	0.98
1q21.1_del	atherosclerotic_vascular_disease	375808	105	5750	1	1.6	0.63	0.93	0.93	0.11	3.37	2942	0.98
2q11.2_del	atherosclerotic_vascular_disease	375732	29	5749	0	0.4	0.00	0.99	1.01	0.01	7.33	3118	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q13_del	biliary	360722	44	20781	7	2.5	2.52	0.0077	3.41	1.44	7.05	102	0.24
16p11.2distal_del	biliary	360725	47	20781	7	2.7	2.38	0.014	3.06	1.29	6.27	145	0.29
10q11.21q11.23_dup	biliary	360713	35	20779	5	2.0	2.30	0.020	3.48	1.25	8.02	184	0.35
15q24_dup	biliary	360684	6	20776	2	0.3	4.59	0.032	6.82	1.22	28.06	251	0.39
1q21.1_dup	biliary	360831	153	20789	15	8.8	1.64	0.055	1.74	0.99	2.86	351	0.49
16p11.2distal_dup	biliary	360794	116	20785	11	6.7	1.59	0.081	1.80	0.92	3.18	454	0.56
PWS_dup	biliary	360692	14	20776	2	0.8	2.30	0.11	3.61	0.71	12.00	550	0.62
17q12_dup	biliary	360769	91	20782	8	5.2	1.48	0.12	1.84	0.84	3.52	596	0.63
TAR_dup	biliary	361058	380	20802	28	21.9	1.26	0.12	1.37	0.92	1.98	600	0.63
15q13.3dup(CHRNA7)	biliary	363349	2671	20946	172	154.0	1.11	0.13	1.13	0.96	1.32	645	0.64
17q12_del	biliary	360684	6	20775	1	0.3	2.62	0.13	5.05	0.52	24.31	652	0.65
16p12.1_del	biliary	360895	217	20792	18	12.5	1.41	0.14	1.46	0.88	2.29	667	0.66
2q13_dup	biliary	360748	70	20775	1	4.0	0.26	0.18	0.39	0.04	1.44	796	0.73
16p11.2_dup	biliary	360799	121	20784	10	7.0	1.40	0.21	1.54	0.77	2.77	872	0.75
16p13.11_del	biliary	360792	114	20784	10	6.6	1.48	0.21	1.53	0.76	2.76	884	0.76
22q11.2_del	biliary	360687	9	20775	1	0.5	1.84	0.23	3.47	0.37	15.31	917	0.77
15q13.3_dup	biliary	360887	209	20789	15	12.0	1.23	0.23	1.40	0.80	2.28	922	0.77
15q13.3del(CHRNA7)	biliary	360687	9	20775	1	0.5	1.84	0.24	3.30	0.35	14.68	973	0.78
3q29_del	biliary	360686	8	20775	1	0.5	2.04	0.25	3.24	0.34	14.83	1006	0.78
17q11.2_del_NF1	biliary	360686	8	20775	1	0.5	2.04	0.28	3.01	0.32	13.64	1081	0.80
NRXN1_del	biliary	360830	152	20779	5	8.8	0.58	0.31	0.66	0.25	1.40	1180	0.81
16p13.11_dup	biliary	361425	747	20810	36	43.0	0.84	0.33	0.85	0.60	1.17	1261	0.82
2q21.1_dup	biliary	360729	51	20778	4	2.9	1.34	0.34	1.66	0.54	3.93	1290	0.83
2q11.2_dup	biliary	360704	26	20774	0	1.5	0.00	0.35	0.33	0.00	2.37	1307	0.83
15q13.3_del	biliary	360712	34	20777	3	2.0	1.49	0.36	1.74	0.47	4.63	1342	0.84
15q11q13del_BP3-BP4	biliary	360691	13	20775	1	0.7	1.31	0.42	2.18	0.24	9.13	1507	0.87
2q11.2_del	biliary	360705	27	20776	2	1.6	1.27	0.42	1.78	0.36	5.48	1526	0.87
22q11.2_dup	biliary	360928	250	20790	16	14.4	1.10	0.48	1.20	0.70	1.92	1718	0.88
15q11q13dup_BP3_BP4	biliary	360725	47	20777	3	2.7	1.10	0.52	1.47	0.40	3.83	1806	0.90
13q12.12_dup	biliary	360881	203	20783	9	11.7	0.78	0.53	0.81	0.39	1.48	1833	0.90
10q11.21q11.23_del	biliary	360730	52	20778	4	3.0	1.31	0.56	1.35	0.44	3.21	1915	0.91
16p12.1_dup	biliary	360858	180	20786	12	10.4	1.15	0.58	1.18	0.63	2.02	1952	0.93
3q29_dup	biliary	360683	5	20774	0	0.29	0.00	0.58	2.54	0.02	24.48	1961	0.93
2q13del(NPHP1)	biliary	362886	2208	20896	122	127.1	0.96	0.62	0.96	0.79	1.14	2075	0.94
TAR_del	biliary	360743	65	20778	4	3.7	1.06	0.64	1.27	0.42	2.97	2112	0.94
17p12_CMT1A_dup	biliary	360787	109	20781	7	6.3	1.11	0.64	1.20	0.52	2.36	2116	0.94
22q11.2distal_dup	biliary	360690	12	20774	0	0.7	0.00	0.68	0.58	0.00	4.52	2237	0.95
2q13dup(NPHP1)	biliary	362450	1772	20870	96	102.0	0.94	0.71	0.96	0.78	1.18	2314	0.96
15q11.2_dup	biliary	362494	1816	20875	101	104.6	0.97	0.74	0.97	0.79	1.18	2403	0.96
WBS_dup	biliary	360691	13	20774	0	0.7	0.00	0.75	0.65	0.01	5.06	2444	0.96
8p23.1_dup	biliary	360683	5	20774	0	0.3	0.00	0.76	1.62	0.01	14.75	2466	0.97
10q23_dup	biliary	360685	7	20774	0	0.4	0.00	0.78	1.54	0.01	13.17	2538	0.97
13q12dup_CRYL1	biliary	360688	10	20774	0	0.6	0.00	0.80	0.71	0.01	5.57	2589	0.97
Potocki_Lupski	biliary	360683	5	20774	0	0.3	0.00	0.81	1.44	0.01	12.96	2626	0.97
22q11.2distal_del	biliary	360683	5	20774	0	0.3	0.00	0.83	1.42	0.01	13.58	2660	0.97
15q11.2_del	biliary	362163	1485	20859	85	85.5	0.99	0.83	1.02	0.82	1.27	2673	0.97
7q11.23_dup_distal	biliary	360700	22	20775	1	1.3	0.80	0.84	1.19	0.13	4.65	2720	0.97
17p12_HNPP_del	biliary	360886	208	20785	11	12.0	0.92	0.85	0.95	0.49	1.64	2745	0.97
2q21.1_del	biliary	360716	38	20776	2	2.2	0.92	0.86	1.12	0.23	3.36	2773	0.98
1q21.1_del	biliary	360779	101	20779	5	5.8	0.87	0.89	0.94	0.35	2.03	2834	0.98
13q12.12_del	biliary	360758	80	20778	4	4.6	0.87	0.91	0.95	0.31	2.20	2910	0.98
16p11.2_del	biliary	360776	98	20779	5	5.6	0.89	0.94	1.03	0.38	2.24	2974	0.99
15q11q13dup_BP3_BP5	biliary	360687	9	20774	0	0.5	0.00	0.95	0.91	0.01	7.40	3000	0.99
13q12del_CRYL1	biliary	361023	345	20792	18	19.9	0.91	0.97	0.99	0.60	1.54	3067	0.99



CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
1q21.1_del	cataract	359694	92	21864	14	5.6	2.31	0.00047	3.23	1.74	5.60	35	0.042*
TAR_dup	cataract	359975	373	21885	35	22.7	1.50	0.0058	1.71	1.18	2.40	84	0.22
17q12_del	cataract	359607	5	21852	2	0.3	4.99	0.014	10.31	1.73	47.81	149	0.30
2q13_del	cataract	359647	45	21856	6	2.7	2.05	0.015	3.41	1.31	7.61	155	0.30
3q29_del	cataract	359609	7	21852	2	0.4	3.88	0.029	7.29	1.28	29.95	239	0.37
16p11.2_del	cataract	359697	95	21858	8	5.8	1.36	0.064	2.14	0.95	4.23	377	0.53
WBS_dup	cataract	359613	11	21852	2	0.7	2.69	0.079	4.45	0.82	16.99	445	0.55
13q12.12_dup	cataract	359797	195	21867	17	11.9	1.40	0.084	1.60	0.94	2.58	466	0.56
16p12.1_dup	cataract	359778	176	21866	16	10.7	1.45	0.090	1.61	0.92	2.64	488	0.58
2q11.2_dup	cataract	359625	23	21853	3	1.4	2.01	0.11	2.92	0.75	8.52	557	0.62
22q11.2distal_dup	cataract	359612	10	21852	2	0.6	2.91	0.13	3.45	0.65	12.47	635	0.64
10q23_dup	cataract	359608	6	21851	1	0.4	2.49	0.13	5.70	0.53	34.62	638	0.64
2q13del(NPHP1)	cataract	361782	2180	22000	150	132.6	1.12	0.15	1.13	0.95	1.34	702	0.69
16p11.2distal_del	cataract	359652	50	21854	4	3.0	1.29	0.20	2.03	0.65	4.92	833	0.75
15q11.2_dup	cataract	361423	1821	21946	96	110.6	0.87	0.28	0.89	0.72	1.09	1091	0.80
15q11q13del_BP3-BP4	cataract	359615	13	21851	1	0.8	1.25	0.30	2.84	0.30	12.45	1150	0.81
16p13.11_del	cataract	359717	115	21859	9	7.0	1.27	0.32	1.44	0.68	2.70	1225	0.81
PWS_dup	cataract	359617	15	21851	1	0.9	1.09	0.33	2.78	0.28	14.05	1244	0.82
15q13.3_del	cataract	359636	34	21853	3	2.1	1.42	0.33	1.83	0.49	5.01	1255	0.82
2q11.2_del	cataract	359629	27	21852	2	1.6	1.20	0.34	2.04	0.40	6.59	1284	0.82
2q13_dup	cataract	359671	69	21852	2	4.2	0.49	0.47	0.64	0.13	1.91	1676	0.88
TAR_del	cataract	359669	67	21852	2	4.1	0.51	0.49	0.65	0.13	1.94	1725	0.88
22q11.2_dup	cataract	359853	251	21865	15	15.3	0.98	0.51	1.20	0.68	1.96	1788	0.89
10q11.21q11.23_dup	cataract	359640	38	21852	2	2.3	0.87	0.51	1.60	0.32	5.04	1798	0.90
2q21.1_del	cataract	359639	37	21853	3	2.2	1.31	0.52	1.48	0.39	4.06	1819	0.90
3q29_dup	cataract	359607	5	21850	0	0.30	0.00	0.54	2.94	0.02	33.18	1855	0.91
10q11.21q11.23_del	cataract	359656	54	21852	2	3.3	0.62	0.55	0.69	0.14	2.07	1884	0.91
16p12.1_del	cataract	359822	220	21865	15	13.4	1.11	0.55	1.18	0.67	1.93	1893	0.92
22q11.2distal_del	cataract	359607	5	21850	0	0.3	0.00	0.61	2.41	0.02	26.87	2036	0.94
1q21.1_dup	cataract	359760	158	21860	10	9.6	1.04	0.63	1.18	0.58	2.12	2084	0.94
17q12_dup	cataract	359695	93	21856	6	5.7	1.06	0.67	1.20	0.48	2.52	2197	0.95
16p11.2_dup	cataract	359725	123	21858	8	7.5	1.07	0.73	1.13	0.52	2.18	2388	0.96
17q11.2_del_NF1	cataract	359611	9	21850	0	0.5	0.00	0.74	0.63	0.00	5.22	2402	0.96
NRXN1_del	cataract	359752	150	21857	7	9.1	0.78	0.74	0.88	0.38	1.74	2405	0.96
15q24_dup	cataract	359610	8	21850	0	0.5	0.00	0.75	1.68	0.01	15.38	2418	0.97
2q21.1_dup	cataract	359655	53	21852	2	3.2	0.63	0.77	0.83	0.17	2.49	2498	0.97
22q11.2_del	cataract	359612	10	21850	0	0.6	0.00	0.79	1.53	0.01	13.51	2554	0.97
16p11.2distal_dup	cataract	359724	122	21855	5	7.4	0.69	0.80	0.89	0.33	1.95	2573	0.97
16p13.11_dup	cataract	360341	739	21894	44	44.9	0.98	0.80	1.04	0.75	1.40	2578	0.97
2q13dup(NPHP1)	cataract	361361	1759	21959	109	106.9	1.02	0.81	1.02	0.84	1.24	2611	0.97
Potocki_Lupski	cataract	359607	5	21850	0	0.3	0.00	0.82	1.44	0.01	14.22	2630	0.97
15q11.2_del	cataract	361080	1478	21942	92	89.8	1.02	0.85	1.02	0.82	1.26	2723	0.97
7q11.23_dup_distal	cataract	359624	22	21851	1	1.3	0.76	0.87	1.16	0.13	4.76	2779	0.98
8p23.1_dup	cataract	359607	5	21850	0	0.3	0.00	0.89	1.24	0.01	11.77	2843	0.98
15q11q13dup_BP3_BP4	cataract	359650	48	21852	2	2.9	0.70	0.90	0.92	0.19	2.82	2864	0.98
13q12dup_CRYL1	cataract	359612	10	21850	0	0.6	0.00	0.91	0.85	0.01	7.08	2901	0.98
13q12.12_del	cataract	359681	79	21855	5	4.8	1.04	0.92	1.05	0.38	2.32	2933	0.99
13q12del_CRYL1	cataract	359947	345	21868	18	21.0	0.87	0.92	1.02	0.61	1.60	2934	0.98
15q13.3dup(CHRNA7)	cataract	362287	2685	22008	158	163.1	0.97	0.93	0.99	0.84	1.17	2953	0.98
17p12_HNPP_del	cataract	359808	206	21863	13	12.5	1.04	0.93	0.98	0.53	1.65	2961	0.99
15q13.3_dup	cataract	359815	213	21861	11	12.9	0.86	0.94	0.98	0.50	1.71	2966	0.99
15q11q13dup_BP3_BP5	cataract	359611	9	21850	0	0.5	0.00	0.95	1.10	0.01	10.14	3010	0.99
15q13.3del(CHRNA7)	cataract	359612	10	21850	0	0.6	0.00	0.97	1.06	0.01	9.26	3055	0.99
17p12_CMT1A_dup	cataract	359712	110	21856	6	6.7	0.90	0.98	0.99	0.40	2.06	3094	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q11.2_dup	cerebrovascular_disease_not_stroke	377146	24	4332	2	0.3	6.78	0.0088	10.31	2.05	32.80	110	0.25
8p23.1_dup	cerebrovascular_disease_not_stroke	377126	4	4331	1	0.0	17.62	0.012	25.97	2.52	148.08	134	0.27
10q23_dup	cerebrovascular_disease_not_stroke	377128	6	4331	1	0.1	12.59	0.013	24.47	2.35	137.34	143	0.28
17q12_del	cerebrovascular_disease_not_stroke	377128	6	4331	1	0.1	12.59	0.017	20.09	2.00	105.62	168	0.31
PWS_dup	cerebrovascular_disease_not_stroke	377137	15	4331	1	0.2	5.51	0.027	14.24	1.48	64.03	229	0.37
22q11.2distal_dup	cerebrovascular_disease_not_stroke	377133	11	4331	1	0.1	7.34	0.046	9.84	1.05	42.62	317	0.46
TAR_del	cerebrovascular_disease_not_stroke	377189	67	4332	2	0.8	2.55	0.11	3.42	0.70	10.08	555	0.63
16p12.1_del	cerebrovascular_disease_not_stroke	377352	230	4335	5	2.6	1.87	0.11	2.16	0.81	4.59	567	0.63
15q13.3dup(CHRNA7)	cerebrovascular_disease_not_stroke	379925	2803	4370	40	32.2	1.24	0.12	1.30	0.93	1.75	591	0.63
16p13.11_del	cerebrovascular_disease_not_stroke	377243	121	4333	3	1.4	2.13	0.12	2.63	0.73	6.65	614	0.63
2q11.2_del	cerebrovascular_disease_not_stroke	377150	28	4331	1	0.3	3.04	0.13	5.00	0.55	19.65	618	0.63
3q29_dup	cerebrovascular_disease_not_stroke	377127	5	4330	0	0.06	0.00	0.19	14.76	0.11	138.15	810	0.74
22q11.2distal_del	cerebrovascular_disease_not_stroke	377127	5	4330	0	0.1	0.00	0.22	11.53	0.09	112.07	915	0.77
2q21.1_del	cerebrovascular_disease_not_stroke	377161	39	4331	1	0.4	2.20	0.24	3.17	0.36	12.05	969	0.78
15q24_dup	cerebrovascular_disease_not_stroke	377130	8	4330	0	0.1	0.00	0.26	9.22	0.07	79.60	1015	0.79
2q13_del	cerebrovascular_disease_not_stroke	377172	50	4331	1	0.6	1.73	0.26	2.99	0.34	11.25	1033	0.79
Potocki_Lupski	cerebrovascular_disease_not_stroke	377127	5	4330	0	0.1	0.00	0.28	8.28	0.06	77.75	1077	0.80
17p12_CMT1A_dup	cerebrovascular_disease_not_stroke	377236	114	4332	2	1.3	1.52	0.34	1.98	0.41	5.71	1274	0.83
22q11.2_del	cerebrovascular_disease_not_stroke	377132	10	4330	0	0.1	0.00	0.34	5.95	0.05	50.34	1280	0.82
15q11q13dup_BP3_BP5	cerebrovascular_disease_not_stroke	377131	9	4330	0	0.1	0.00	0.36	5.44	0.04	46.94	1336	0.84
2q13dup(NPHP1)	cerebrovascular_disease_not_stroke	378973	1851	4347	17	21.2	0.80	0.37	0.81	0.49	1.26	1361	0.85
10q11.21q11.23_del	cerebrovascular_disease_not_stroke	377177	55	4331	1	0.6	1.57	0.37	2.32	0.26	8.68	1375	0.85
15q13.3del(CHRNA7)	cerebrovascular_disease_not_stroke	377132	10	4330	0	0.1	0.00	0.38	4.89	0.04	39.47	1405	0.85
3q29_del	cerebrovascular_disease_not_stroke	377131	9	4330	0	0.1	0.00	0.38	4.86	0.04	40.35	1415	0.85
16p12.1_dup	cerebrovascular_disease_not_stroke	377311	189	4333	3	2.2	1.38	0.38	1.66	0.46	4.15	1419	0.85
13q12dup_CRYL1	cerebrovascular_disease_not_stroke	377132	10	4330	0	0.1	0.00	0.40	4.56	0.04	36.95	1466	0.85
15q11q13del_BP3-BP4	cerebrovascular_disease_not_stroke	377136	14	4330	0	0.2	0.00	0.42	4.15	0.03	31.64	1517	0.87
16p11.2_dup	cerebrovascular_disease_not_stroke	377251	129	4332	2	1.5	1.34	0.47	1.65	0.34	4.73	1675	0.88
WBS_dup	cerebrovascular_disease_not_stroke	377135	13	4330	0	0.1	0.00	0.48	3.40	0.03	26.91	1697	0.88
17q11.2_del_NF1	cerebrovascular_disease_not_stroke	377131	9	4330	0	0.1	0.00	0.48	3.37	0.03	27.49	1708	0.88
13q12del_CRYL1	cerebrovascular_disease_not_stroke	377483	361	4332	2	4.1	0.49	0.51	0.68	0.14	1.91	1791	0.89
1q21.1_del	cerebrovascular_disease_not_stroke	377228	106	4330	0	1.2	0.00	0.51	0.45	0.00	3.08	1794	0.90
16p11.2_del	cerebrovascular_disease_not_stroke	377225	103	4330	0	1.2	0.00	0.56	0.48	0.00	3.32	1906	0.91
15q13.3_dup	cerebrovascular_disease_not_stroke	377343	221	4333	3	2.5	1.18	0.58	1.37	0.38	3.40	1958	0.93
17p12_HNPP_del	cerebrovascular_disease_not_stroke	377338	216	4333	3	2.5	1.21	0.62	1.33	0.37	3.30	2056	0.94
7q11.23_dup_distal	cerebrovascular_disease_not_stroke	377145	23	4330	0	0.3	0.00	0.66	2.04	0.02	15.06	2166	0.95
22q11.2_dup	cerebrovascular_disease_not_stroke	377385	263	4333	3	3.0	0.99	0.68	1.26	0.35	3.12	2231	0.95
15q11.2_del	cerebrovascular_disease_not_stroke	378676	1554	4346	16	17.8	0.90	0.70	0.91	0.54	1.43	2282	0.96
13q12.12_del	cerebrovascular_disease_not_stroke	377205	83	4331	1	1.0	1.05	0.70	1.41	0.16	5.16	2284	0.95
16p13.11_dup	cerebrovascular_disease_not_stroke	377898	776	4337	7	8.9	0.79	0.71	0.87	0.39	1.67	2305	0.96
2q13_dup	cerebrovascular_disease_not_stroke	377193	71	4330	0	0.8	0.00	0.72	0.63	0.00	4.37	2362	0.96
17q12_dup	cerebrovascular_disease_not_stroke	377220	98	4331	1	1.1	0.89	0.78	1.27	0.14	4.62	2535	0.97
1q21.1_dup	cerebrovascular_disease_not_stroke	377289	167	4331	1	1.9	0.52	0.78	0.80	0.09	2.90	2541	0.97
10q11.21q11.23_dup	cerebrovascular_disease_not_stroke	377162	40	4330	0	0.5	0.00	0.82	1.42	0.01	10.15	2632	0.97
16p11.2distal_dup	cerebrovascular_disease_not_stroke	377248	126	4331	1	1.4	0.69	0.85	1.18	0.13	4.28	2724	0.97
2q13del(NPHP1)	cerebrovascular_disease_not_stroke	379425	2303	4357	27	26.4	1.02	0.85	1.04	0.69	1.48	2728	0.97
2q21.1_dup	cerebrovascular_disease_not_stroke	377177	55	4330	0	0.6	0.00	0.86	0.79	0.01	5.55	2776	0.98
NRXN1_del	cerebrovascular_disease_not_stroke	377278	156	4331	1	1.8	0.56	0.87	0.88	0.10	3.17	2781	0.98
13q12.12_dup	cerebrovascular_disease_not_stroke	377332	210	4332	2	2.4	0.83	0.89	1.09	0.23	3.10	2849	0.98
16p11.2distal_del	cerebrovascular_disease_not_stroke	377176	54	4330	0	0.6	0.00	0.93	0.88	0.01	6.17	2951	0.98
15q11q13dup_BP3_BP4	cerebrovascular_disease_not_stroke	377172	50	4330	0	0.6	0.00	0.95	0.92	0.01	6.48	3015	0.99
15q11.2_dup	cerebrovascular_disease_not_stroke	379018	1896	4351	21	21.8	0.97	0.97	1.01	0.64	1.51	3054	0.99
15q13.3_del	cerebrovascular_disease_not_stroke	377159	37	4330	0	0.4	0.00	0.97	1.06	0.01	7.60	3057	0.99
TAR_dup	cerebrovascular_disease_not_stroke	377526	404	4334	4	4.6	0.86	0.99	1.01	0.34	2.27	3096	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
17q12_del	coagulation_defects	378917	6	2542	1	0.0	21.45	0.0089	28.68	2.96	138.21	112	0.25
15q11q13del_BP3-BP4	coagulation_defects	378924	13	2542	1	0.1	10.72	0.018	17.53	1.91	71.93	172	0.32
2q13_del	coagulation_defects	378960	49	2543	2	0.3	5.89	0.019	7.24	1.49	21.27	175	0.34
PWS_dup	coagulation_defects	378926	15	2542	1	0.1	9.38	0.022	15.33	1.68	61.92	199	0.34
16p11.2_del	coagulation_defects	379011	100	2544	3	0.7	4.37	0.024	4.60	1.28	11.58	210	0.35
22q11.2_dup	coagulation_defects	379172	261	2546	5	1.8	2.82	0.031	2.98	1.12	6.30	248	0.39
17q12_dup	coagulation_defects	379008	97	2543	2	0.7	3.03	0.096	3.62	0.75	10.39	508	0.59
16p11.2distal_dup	coagulation_defects	379036	125	2543	2	0.8	2.36	0.15	2.93	0.61	8.36	696	0.68
16p11.2distal_del	coagulation_defects	378964	53	2542	1	0.4	2.78	0.17	3.96	0.45	14.57	754	0.72
3q29_dup	coagulation_defects	378916	5	2541	0	0.03	0.00	0.19	15.10	0.11	135.65	802	0.73
8p23.1_dup	coagulation_defects	378916	5	2541	0	0.0	0.00	0.20	13.44	0.10	119.18	847	0.74
Potocki_Lupski	coagulation_defects	378916	5	2541	0	0.0	0.00	0.21	13.00	0.10	115.16	864	0.74
22q11.2distal_del	coagulation_defects	378916	5	2541	0	0.0	0.00	0.23	11.02	0.08	99.34	928	0.77
2q13_dup	coagulation_defects	378981	70	2542	1	0.5	2.11	0.24	3.16	0.36	11.54	962	0.78
15q24_dup	coagulation_defects	378919	8	2541	0	0.1	0.00	0.25	9.62	0.07	77.57	989	0.78
10q23_dup	coagulation_defects	378918	7	2541	0	0.0	0.00	0.25	9.63	0.07	80.51	992	0.78
2q13dup(NPHP1)	coagulation_defects	380762	1851	2558	17	12.4	1.37	0.28	1.32	0.78	2.07	1102	0.80
15q11q13dup_BP3_BP5	coagulation_defects	378920	9	2541	0	0.1	0.00	0.28	7.68	0.06	60.77	1113	0.80
3q29_del	coagulation_defects	378920	9	2541	0	0.1	0.00	0.29	7.39	0.06	58.44	1128	0.81
17q11.2_del_NF1	coagulation_defects	378920	9	2541	0	0.1	0.00	0.29	7.26	0.06	57.46	1138	0.81
13q12dup_CRYL1	coagulation_defects	378921	10	2541	0	0.1	0.00	0.30	7.17	0.06	55.75	1144	0.81
13q12.12_del	coagulation_defects	378994	83	2542	1	0.6	1.79	0.30	2.71	0.31	9.84	1148	0.81
15q13.3_dup	coagulation_defects	379135	224	2541	0	1.5	0.00	0.31	0.31	0.00	2.11	1185	0.81
22q11.2_del	coagulation_defects	378921	10	2541	0	0.1	0.00	0.32	6.51	0.05	51.00	1213	0.81
15q13.3del(CHRNA7)	coagulation_defects	378921	10	2541	0	0.1	0.00	0.32	6.37	0.05	49.77	1229	0.81
16p12.1_dup	coagulation_defects	379101	190	2543	2	1.3	1.56	0.34	1.97	0.41	5.57	1288	0.82
22q11.2distal_dup	coagulation_defects	378923	12	2541	0	0.1	0.00	0.36	5.22	0.04	39.79	1344	0.84
WBS_dup	coagulation_defects	378924	13	2541	0	0.1	0.00	0.38	4.84	0.04	36.78	1407	0.85
13q12.12_dup	coagulation_defects	379121	210	2543	2	1.4	1.42	0.42	1.76	0.37	4.99	1505	0.87
1q21.1_del	coagulation_defects	379016	105	2542	1	0.7	1.42	0.44	2.05	0.23	7.41	1561	0.87
7q11.23_dup_distal	coagulation_defects	378934	23	2541	0	0.2	0.00	0.49	3.24	0.03	23.30	1728	0.89
17p12_CMT1A_dup	coagulation_defects	379026	115	2542	1	0.8	1.29	0.49	1.87	0.21	6.74	1735	0.89
16p12.1_del	coagulation_defects	379144	233	2543	2	1.6	1.28	0.52	1.55	0.33	4.40	1807	0.90
2q11.2_dup	coagulation_defects	378937	26	2541	0	0.2	0.00	0.52	2.92	0.02	20.85	1823	0.90
NRXN1_del	coagulation_defects	379068	157	2541	0	1.1	0.00	0.54	0.47	0.00	3.21	1869	0.91
13q12del_CRYL1	coagulation_defects	379273	362	2542	1	2.4	0.41	0.56	0.64	0.07	2.29	1913	0.91
15q11.2_del	coagulation_defects	380469	1558	2553	12	10.5	1.15	0.58	1.18	0.64	1.96	1950	0.93
16p11.2_dup	coagulation_defects	379041	130	2542	1	0.9	1.15	0.58	1.62	0.19	5.85	1972	0.93
2q11.2_del	coagulation_defects	378940	29	2541	0	0.2	0.00	0.59	2.42	0.02	17.21	1983	0.93
2q13del(NPHP1)	coagulation_defects	381224	2313	2558	17	15.5	1.10	0.63	1.13	0.68	1.75	2085	0.94
TAR_dup	coagulation_defects	379316	405	2544	3	2.7	1.10	0.66	1.27	0.36	3.13	2188	0.95
10q11.21q11.23_dup	coagulation_defects	378951	40	2541	0	0.3	0.00	0.68	1.91	0.02	13.38	2242	0.95
16p13.11_del	coagulation_defects	379035	124	2541	0	0.8	0.00	0.69	0.60	0.00	4.07	2267	0.95
16p13.11_dup	coagulation_defects	379690	779	2545	4	5.2	0.77	0.72	0.85	0.28	1.90	2334	0.96
2q21.1_del	coagulation_defects	378951	40	2541	0	0.3	0.00	0.72	1.74	0.01	12.20	2348	0.96
15q13.3_del	coagulation_defects	378948	37	2541	0	0.2	0.00	0.72	1.73	0.01	12.20	2359	0.96
1q21.1_dup	coagulation_defects	379078	167	2542	1	1.1	0.89	0.78	1.27	0.15	4.57	2518	0.97
15q11q13dup_BP3_BP4	coagulation_defects	378961	50	2541	0	0.3	0.00	0.80	1.45	0.01	10.12	2595	0.97
2q21.1_dup	coagulation_defects	378966	55	2541	0	0.4	0.00	0.86	1.31	0.01	9.10	2754	0.97
15q13.3dup(CHRNA7)	coagulation_defects	381735	2824	2560	19	18.9	1.00	0.87	1.04	0.64	1.58	2787	0.98
15q11.2_dup	coagulation_defects	380815	1904	2554	13	12.8	1.02	0.87	1.05	0.58	1.71	2792	0.98
10q11.21q11.23_del	coagulation_defects	378967	56	2541	0	0.4	0.00	0.89	1.22	0.01	8.49	2846	0.98
TAR_del	coagulation_defects	378980	69	2541	0	0.5	0.00	0.95	1.09	0.01	7.55	3013	0.99
17p12_HNPP_del	coagulation_defects	379129	218	2542	1	1.5	0.69	0.98	1.02	0.12	3.65	3090	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
1q21.1_del	congenital	376526	101	5032	5	1.3	3.58	0.0098	3.92	1.46	8.42	122	0.25
10q23_dup	congenital	376431	6	5028	1	0.1	10.84	0.021	16.58	1.72	79.30	190	0.35
17q11.2_del_NF1	congenital	376433	8	5028	1	0.1	8.43	0.027	14.03	1.49	62.26	225	0.37
15q11q13dup_BP3_BP5	congenital	376433	8	5028	1	0.1	8.43	0.035	11.81	1.26	52.52	265	0.41
15q13.3_dup	congenital	376642	217	5034	7	2.9	2.37	0.035	2.44	1.07	4.70	269	0.41
15q11q13del_BP3-BP4	congenital	376438	13	5028	1	0.2	5.42	0.062	7.98	0.87	32.71	370	0.53
17p12_HNPP_del	congenital	376644	219	5027	0	2.9	0.00	0.083	0.17	0.00	1.17	464	0.56
7q11.23_dup_distal	congenital	376447	22	5028	1	0.3	3.30	0.12	5.13	0.57	19.86	595	0.63
2q13del(NPHP1)	congenital	378717	2292	5065	38	30.7	1.24	0.18	1.25	0.90	1.70	776	0.72
17p12_CMT1A_dup	congenital	376538	113	5030	3	1.5	1.96	0.19	2.22	0.62	5.56	812	0.74
16p13.11_del	congenital	376546	121	5030	3	1.6	1.84	0.22	2.12	0.59	5.29	891	0.76
2q21.1_del	congenital	376464	39	5028	1	0.5	1.90	0.29	2.76	0.31	10.28	1136	0.81
3q29_dup	congenital	376430	5	5027	0	0.07	0.00	0.29	7.35	0.06	65.15	1140	0.81
8p23.1_dup	congenital	376430	5	5027	0	0.1	0.00	0.31	6.84	0.05	60.46	1186	0.82
16p11.2_del	congenital	376528	103	5027	0	1.4	0.00	0.31	0.31	0.00	2.15	1210	0.81
15q13.3dup(CHRNA7)	congenital	379237	2812	5058	31	37.5	0.83	0.31	0.84	0.58	1.17	1211	0.81
Potocki_Lupski	congenital	376430	5	5027	0	0.1	0.00	0.32	6.32	0.05	55.90	1239	0.82
22q11.2distal_del	congenital	376430	5	5027	0	0.1	0.00	0.35	5.69	0.04	50.48	1309	0.83
17q12_dup	congenital	376522	97	5029	2	1.3	1.53	0.37	1.89	0.39	5.43	1356	0.84
TAR_dup	congenital	376830	405	5030	3	5.4	0.56	0.37	0.64	0.18	1.57	1360	0.85
17q12_del	congenital	376432	7	5027	0	0.1	0.00	0.39	4.66	0.04	38.37	1452	0.85
15q24_dup	congenital	376433	8	5027	0	0.1	0.00	0.42	4.10	0.03	32.99	1533	0.87
3q29_del	congenital	376434	9	5027	0	0.1	0.00	0.46	3.63	0.03	28.64	1632	0.88
16p11.2distal_del	congenital	376478	53	5028	1	0.7	1.41	0.46	1.98	0.22	7.27	1633	0.88
13q12dup_CRYL1	congenital	376435	10	5027	0	0.1	0.00	0.48	3.38	0.03	26.29	1700	0.88
10q11.21q11.23_del	congenital	376480	55	5028	1	0.7	1.36	0.49	1.87	0.21	6.86	1738	0.89
22q11.2_dup	congenital	376689	264	5029	2	3.5	0.57	0.49	0.67	0.14	1.88	1743	0.89
15q13.3del(CHRNA7)	congenital	376435	10	5027	0	0.1	0.00	0.50	3.14	0.02	24.43	1764	0.89
13q12.12_del	congenital	376509	84	5027	0	1.1	0.00	0.50	0.44	0.00	3.02	1770	0.89
16p13.11_dup	congenital	377200	775	5035	8	10.3	0.78	0.52	0.81	0.38	1.48	1815	0.90
22q11.2_del	congenital	376435	10	5027	0	0.1	0.00	0.52	2.96	0.02	23.06	1822	0.90
22q11.2distal_dup	congenital	376437	12	5027	0	0.2	0.00	0.54	2.78	0.02	21.16	1864	0.91
1q21.1_dup	congenital	376592	167	5028	1	2.2	0.45	0.56	0.64	0.07	2.29	1902	0.92
TAR_del	congenital	376493	68	5028	1	0.9	1.10	0.57	1.67	0.19	6.10	1931	0.92
WBS_dup	congenital	376438	13	5027	0	0.2	0.00	0.57	2.57	0.02	19.47	1935	0.92
16p12.1_dup	congenital	376614	189	5030	3	2.5	1.19	0.59	1.36	0.38	3.37	1981	0.93
2q13_dup	congenital	376496	71	5027	0	0.9	0.00	0.60	0.51	0.00	3.54	2008	0.93
16p11.2_dup	congenital	376554	129	5029	2	1.7	1.16	0.62	1.39	0.29	3.96	2073	0.94
PWS_dup	congenital	376441	16	5027	0	0.2	0.00	0.66	2.03	0.02	15.03	2171	0.95
16p12.1_del	congenital	376658	233	5029	2	3.1	0.65	0.67	0.77	0.16	2.18	2217	0.95
13q12.12_dup	congenital	376634	209	5030	3	2.8	1.07	0.70	1.24	0.35	3.05	2303	0.96
13q12del_CRYL1	congenital	376783	358	5032	5	4.8	1.05	0.74	1.16	0.44	2.43	2400	0.96
2q21.1_dup	congenital	376480	55	5027	0	0.7	0.00	0.75	0.66	0.01	4.54	2426	0.97
2q13_del	congenital	376476	51	5027	0	0.7	0.00	0.77	0.68	0.01	4.74	2507	0.97
NRXN1_del	congenital	376580	155	5029	2	2.1	0.97	0.79	1.19	0.25	3.38	2557	0.97
15q11q13dup_BP3_BP4	congenital	376475	50	5027	0	0.7	0.00	0.81	0.73	0.01	5.04	2615	0.97
2q11.2_dup	congenital	376451	26	5027	0	0.3	0.00	0.83	1.39	0.01	9.92	2665	0.97
15q11.2_dup	congenital	378318	1893	5051	24	25.3	0.95	0.84	0.96	0.63	1.40	2705	0.97
16p11.2distal_dup	congenital	376551	126	5028	1	1.7	0.60	0.84	0.85	0.10	3.08	2719	0.97
2q13dup(NPHP1)	congenital	378268	1843	5052	25	24.6	1.02	0.88	1.03	0.68	1.49	2812	0.98
15q13.3_del	congenital	376462	37	5027	0	0.5	0.00	0.91	0.86	0.01	6.03	2909	0.98
2q11.2_del	congenital	376454	29	5027	0	0.4	0.00	0.92	1.16	0.01	8.21	2930	0.98
15q11.2_del	congenital	377975	1550	5047	20	20.7	0.97	0.93	0.98	0.61	1.48	2959	0.99
10q11.21q11.23_dup	congenital	376465	40	5027	0	0.5	0.00	0.95	0.92	0.01	6.43	3020	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q13_del	connective_tissue_disorder	372146	46	9357	5	1.2	4.00	0.0022	5.58	2.02	12.58	58	0.12
3q29_dup	connective_tissue_disorder	372104	4	9353	1	0.10	8.16	0.014	24.49	2.23	150.02	150	0.30
16p11.2_del	connective_tissue_disorder	372197	97	9358	6	2.4	2.38	0.029	2.79	1.13	5.76	240	0.38
Potocki_Lupski	connective_tissue_disorder	372104	4	9353	1	0.1	8.16	0.037	12.19	1.21	67.29	275	0.42
17q12_dup	connective_tissue_disorder	372194	94	9357	5	2.4	2.06	0.056	2.62	0.97	5.67	355	0.50
17p12_CMT1A_dup	connective_tissue_disorder	372210	110	9358	6	2.8	2.11	0.059	2.37	0.96	4.87	362	0.51
16p11.2distal_dup	connective_tissue_disorder	372227	127	9352	0	3.2	0.00	0.069	0.16	0.00	1.11	398	0.55
1q21.1_del	connective_tissue_disorder	372201	101	9357	5	2.5	1.92	0.11	2.17	0.80	4.70	574	0.63
WBS_dup	connective_tissue_disorder	372112	12	9353	1	0.3	3.14	0.14	4.71	0.50	20.38	674	0.67
13q12.12_del	connective_tissue_disorder	372180	80	9356	4	2.0	1.94	0.15	2.17	0.71	5.05	703	0.69
16p13.11_del	connective_tissue_disorder	372219	119	9357	5	3.0	1.64	0.23	1.78	0.66	3.82	924	0.77
15q13.3_dup	connective_tissue_disorder	372317	217	9359	7	5.5	1.27	0.33	1.47	0.64	2.84	1266	0.82
2q11.2_del	connective_tissue_disorder	372128	28	9353	1	0.7	1.41	0.36	2.39	0.27	9.22	1341	0.84
NRXN1_del	connective_tissue_disorder	372252	152	9357	5	3.8	1.30	0.38	1.51	0.56	3.22	1387	0.85
16p11.2distal_del	connective_tissue_disorder	372154	54	9352	0	1.4	0.00	0.45	0.40	0.00	2.77	1600	0.87
8p23.1_dup	connective_tissue_disorder	372105	5	9352	0	0.1	0.00	0.45	3.74	0.03	34.00	1624	0.88
10q23_dup	connective_tissue_disorder	372107	7	9352	0	0.2	0.00	0.47	3.55	0.03	30.79	1664	0.88
10q11.21q11.23_del	connective_tissue_disorder	372154	54	9354	2	1.4	1.46	0.48	1.64	0.34	4.83	1706	0.88
17q12_del	connective_tissue_disorder	372107	7	9352	0	0.2	0.00	0.49	3.25	0.02	26.96	1739	0.89
TAR_del	connective_tissue_disorder	372167	67	9354	2	1.7	1.18	0.50	1.59	0.33	4.65	1762	0.89
16p11.2_dup	connective_tissue_disorder	372227	127	9356	4	3.2	1.25	0.51	1.40	0.46	3.20	1785	0.89
2q13del(NPHP1)	connective_tissue_disorder	374368	2268	9414	62	57.0	1.09	0.51	1.09	0.84	1.39	1792	0.89
22q11.2distal_del	connective_tissue_disorder	372105	5	9352	0	0.1	0.00	0.52	3.07	0.02	30.40	1812	0.90
2q13_dup	connective_tissue_disorder	372169	69	9354	2	1.7	1.15	0.52	1.56	0.32	4.55	1816	0.90
16p12.1_dup	connective_tissue_disorder	372289	189	9355	3	4.7	0.64	0.53	0.72	0.20	1.79	1832	0.90
1q21.1_dup	connective_tissue_disorder	372263	163	9357	5	4.1	1.21	0.55	1.32	0.49	2.80	1876	0.91
15q11.2_del	connective_tissue_disorder	373629	1529	9393	41	38.4	1.07	0.55	1.10	0.79	1.48	1890	0.92
13q12.12_dup	connective_tissue_disorder	372306	206	9358	6	5.2	1.15	0.57	1.27	0.52	2.56	1937	0.92
22q11.2_del	connective_tissue_disorder	372110	10	9352	0	0.3	0.00	0.59	2.45	0.02	19.50	1982	0.93
15q24_dup	connective_tissue_disorder	372108	8	9352	0	0.2	0.00	0.60	2.40	0.02	19.93	2004	0.93
16p13.11_dup	connective_tissue_disorder	372862	762	9373	21	19.2	1.09	0.61	1.12	0.71	1.68	2022	0.94
2q13dup(NPHP1)	connective_tissue_disorder	373920	1820	9400	48	45.8	1.05	0.61	1.08	0.80	1.42	2034	0.94
15q13.3del(CHRNA7)	connective_tissue_disorder	372110	10	9352	0	0.3	0.00	0.62	2.22	0.02	17.79	2074	0.94
15q13.3_del	connective_tissue_disorder	372136	36	9353	1	0.9	1.10	0.63	1.54	0.17	5.85	2091	0.94
3q29_del	connective_tissue_disorder	372109	9	9352	0	0.2	0.00	0.64	2.13	0.02	17.25	2127	0.95
15q11q13dup_BP3_BP5	connective_tissue_disorder	372109	9	9352	0	0.2	0.00	0.65	2.10	0.02	17.28	2151	0.94
2q21.1_del	connective_tissue_disorder	372139	39	9353	1	1.0	1.02	0.66	1.48	0.17	5.61	2167	0.95
17q11.2_del_NF1	connective_tissue_disorder	372109	9	9352	0	0.2	0.00	0.67	1.98	0.02	15.86	2209	0.95
17p12_HNPP_del	connective_tissue_disorder	372313	213	9358	6	5.4	1.12	0.69	1.18	0.48	2.38	2266	0.95
10q11.21q11.23_dup	connective_tissue_disorder	372140	40	9352	0	1.0	0.00	0.72	0.62	0.00	4.41	2336	0.96
15q11q13dup_BP3_BP4	connective_tissue_disorder	372149	49	9353	1	1.2	0.82	0.73	1.35	0.15	5.06	2383	0.96
13q12dup_CRYL1	connective_tissue_disorder	372110	10	9352	0	0.3	0.00	0.75	1.64	0.01	12.98	2435	0.97
15q11q13del_BP3-BP4	connective_tissue_disorder	372114	14	9352	0	0.4	0.00	0.76	1.61	0.01	12.28	2462	0.97
15q11.2_dup	connective_tissue_disorder	373972	1872	9397	45	47.0	0.96	0.78	0.96	0.70	1.27	2519	0.97
PWS_dup	connective_tissue_disorder	372116	16	9352	0	0.4	0.00	0.78	1.53	0.01	11.51	2539	0.97
16p12.1_del	connective_tissue_disorder	372329	229	9358	6	5.8	1.04	0.79	1.11	0.46	2.24	2569	0.97
13q12del_CRYL1	connective_tissue_disorder	372456	356	9359	7	8.9	0.79	0.81	0.91	0.40	1.76	2604	0.97
2q21.1_dup	connective_tissue_disorder	372154	54	9353	1	1.4	0.74	0.84	1.19	0.13	4.42	2684	0.97
2q11.2_dup	connective_tissue_disorder	372126	26	9352	0	0.7	0.00	0.85	0.78	0.01	5.65	2751	0.97
15q13.3dup(CHRNA7)	connective_tissue_disorder	374876	2776	9419	67	69.7	0.96	0.86	0.98	0.76	1.24	2758	0.97
22q11.2distal_dup	connective_tissue_disorder	372112	12	9352	0	0.3	0.00	0.89	1.22	0.01	9.68	2855	0.98
7q11.23_dup_distal	connective_tissue_disorder	372123	23	9352	0	0.6	0.00	0.92	0.87	0.01	6.38	2936	0.99
22q11.2_dup	connective_tissue_disorder	372360	260	9358	6	6.5	0.92	0.95	1.03	0.42	2.06	3003	0.99
TAR_dup	connective_tissue_disorder	372499	399	9361	9	10.0	0.90	0.99	1.00	0.49	1.79	3117	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
PWS_dup	COPD	367237	13	14231	3	0.5	5.03	0.0018	11.84	2.90	37.33	51	0.11
1q21.1_del	COPD	367319	95	14239	11	3.7	2.78	0.0021	3.13	1.57	5.69	56	0.11
TAR_dup	COPD	367606	382	14254	26	14.8	1.71	0.0056	1.86	1.21	2.73	83	0.21
TAR_del	COPD	367286	62	14235	7	2.4	2.72	0.0064	3.54	1.48	7.34	90	0.22
15q13.3_del	COPD	367255	31	14234	6	1.2	4.35	0.0093	3.84	1.44	8.82	120	0.24
16p12.1_del	COPD	367442	218	14245	17	8.5	1.94	0.010	2.04	1.20	3.27	127	0.26
3q29_dup	COPD	367228	4	14229	1	0.15	5.36	0.020	18.81	1.79	112.34	179	0.35
22q11.2_del	COPD	367232	8	14230	2	0.3	5.36	0.025	7.60	1.35	30.44	218	0.36
8p23.1_dup	COPD	367228	4	14229	1	0.2	5.36	0.051	9.97	0.99	54.52	331	0.48
17p12_CMT1A_dup	COPD	367331	107	14237	9	4.1	2.08	0.053	2.11	0.99	3.99	346	0.48
2q21.1_del	COPD	367260	36	14232	4	1.4	2.68	0.072	2.86	0.90	7.15	415	0.54
15q13.3_dup	COPD	367433	209	14243	15	8.1	1.80	0.10	1.61	0.91	2.65	530	0.60
13q12.12_dup	COPD	367424	200	14240	12	7.8	1.52	0.10	1.68	0.89	2.89	533	0.60
16p11.2_del	COPD	367318	94	14237	9	3.6	2.34	0.12	1.83	0.85	3.52	583	0.63
2q13_del	COPD	367271	47	14232	4	1.8	2.10	0.13	2.38	0.75	5.87	627	0.64
16p11.2distal_del	COPD	367278	54	14228	0	2.1	0.00	0.16	0.22	0.00	1.54	727	0.69
15q11q13del_BP3-BP4	COPD	367237	13	14229	1	0.5	1.91	0.17	4.10	0.44	17.46	760	0.72
1q21.1_dup	COPD	367382	158	14238	10	6.1	1.60	0.20	1.55	0.77	2.80	861	0.75
17q12_dup	COPD	367317	93	14234	6	3.6	1.62	0.23	1.71	0.69	3.56	916	0.77
16p12.1_dup	COPD	367406	182	14238	10	7.1	1.40	0.25	1.49	0.74	2.68	994	0.78
15q13.3del(CHRNA7)	COPD	367233	9	14229	1	0.3	2.68	0.26	3.23	0.33	15.36	1024	0.79
13q12.12_del	COPD	367303	79	14233	5	3.1	1.60	0.27	1.70	0.62	3.78	1065	0.80
16p13.11_dup	COPD	367971	747	14264	36	29.0	1.23	0.29	1.21	0.85	1.67	1120	0.80
22q11.2_dup	COPD	367476	252	14242	14	9.8	1.41	0.34	1.32	0.73	2.19	1283	0.82
15q11.2_del	COPD	368741	1517	14281	53	58.8	0.91	0.36	0.88	0.66	1.15	1349	0.85
15q11.2_dup	COPD	369060	1836	14309	81	71.2	1.13	0.37	1.11	0.88	1.38	1383	0.85
2q13_dup	COPD	367294	70	14229	1	2.7	0.38	0.40	0.53	0.06	1.98	1469	0.85
16p11.2distal_dup	COPD	367345	121	14234	6	4.7	1.27	0.42	1.42	0.57	2.94	1512	0.87
NRXN1_del	COPD	367374	150	14235	7	5.8	1.20	0.42	1.38	0.60	2.71	1523	0.87
7q11.23_dup_distal	COPD	367246	22	14229	1	0.9	1.17	0.44	2.06	0.23	8.26	1595	0.87
10q11.21q11.23_dup	COPD	367264	40	14228	0	1.5	0.00	0.46	0.40	0.00	2.92	1637	0.88
15q13.3dup(CHRNA7)	COPD	369972	2748	14323	95	106.4	0.90	0.46	0.93	0.75	1.13	1639	0.88
22q11.2distal_dup	COPD	367235	11	14229	1	0.4	2.23	0.47	2.09	0.20	10.26	1670	0.88
2q11.2_dup	COPD	367249	25	14229	1	1.0	1.03	0.57	1.70	0.19	6.87	1932	0.92
15q24_dup	COPD	367232	8	14228	0	0.3	0.00	0.58	2.48	0.02	20.56	1970	0.93
Potocki_Lupski	COPD	367229	5	14228	0	0.2	0.00	0.59	2.49	0.02	22.84	1976	0.93
2q21.1_dup	COPD	367278	54	14229	1	2.1	0.49	0.64	0.69	0.08	2.60	2133	0.94
2q13dup(NPHP1)	COPD	369020	1796	14300	72	69.6	1.03	0.66	1.06	0.82	1.33	2175	0.95
10q11.21q11.23_del	COPD	367277	53	14231	3	2.1	1.44	0.67	1.29	0.34	3.45	2210	0.95
2q13del(NPHP1)	COPD	369471	2247	14311	83	87.0	0.96	0.72	0.96	0.76	1.19	2351	0.96
10q23_dup	COPD	367231	7	14228	0	0.3	0.00	0.72	1.77	0.01	16.51	2366	0.96
17p12_HNPP_del	COPD	367433	209	14238	10	8.1	1.22	0.77	1.11	0.53	2.03	2493	0.97
15q11q13dup_BP3_BP4	COPD	367273	49	14229	1	1.9	0.54	0.81	0.82	0.09	3.09	2602	0.97
17q12_del	COPD	367231	7	14228	0	0.3	0.00	0.83	1.38	0.01	12.35	2683	0.97
2q11.2_del	COPD	367252	28	14229	1	1.1	0.92	0.84	1.20	0.13	4.76	2689	0.97
15q11q13dup_BP3_BP5	COPD	367233	9	14228	0	0.3	0.00	0.85	1.33	0.01	11.03	2750	0.97
3q29_del	COPD	367233	9	14228	0	0.3	0.00	0.86	1.32	0.01	10.84	2755	0.97
22q11.2distal_del	COPD	367229	5	14228	0	0.2	0.00	0.89	1.25	0.01	15.33	2852	0.98
17q11.2_del_NF1	COPD	367233	9	14228	0	0.3	0.00	0.89	1.22	0.01	9.87	2859	0.98
WBS_dup	COPD	367237	13	14228	0	0.5	0.00	0.89	0.83	0.01	6.54	2862	0.98
13q12dup_CRYL1	COPD	367234	10	14228	0	0.4	0.00	0.92	1.15	0.01	9.32	2940	0.99
16p13.11_del	COPD	367344	120	14232	4	4.6	0.86	0.97	0.98	0.32	2.28	3052	0.99
16p11.2_dup	COPD	367350	126	14233	5	4.9	1.02	0.99	1.00	0.37	2.18	3120	1.00
13q12del_CRYL1	COPD	367576	352	14239	11	13.6	0.81	1.00	1.00	0.52	1.73	3124	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
17q12_del	diabetes_insulin_dependent	378861	3	2598	4	0.0	84.03	3.93E-08	135.87	31.15	641.08	4	3.08E-05*
16p11.2distal_del	diabetes_insulin_dependent	378909	51	2597	3	0.3	8.17	0.0033	8.60	2.35	22.23	66	0.15
Potocki_Lupski	diabetes_insulin_dependent	378862	4	2595	1	0.0	29.41	0.0041	48.85	4.83	270.84	74	0.17
22q11.2distal_del	diabetes_insulin_dependent	378862	4	2595	1	0.0	29.41	0.0063	37.86	3.70	213.72	88	0.22
15q13.3del(CHRNA7)	diabetes_insulin_dependent	378867	9	2595	1	0.1	14.71	0.021	16.19	1.70	72.94	195	0.35
1q21.1_dup	diabetes_insulin_dependent	379022	164	2598	4	1.1	3.50	0.025	3.63	1.20	8.26	220	0.36
16p11.2_del	diabetes_insulin_dependent	378958	100	2597	3	0.7	4.28	0.045	3.75	1.04	9.48	310	0.45
TAR_dup	diabetes_insulin_dependent	379260	402	2600	6	2.8	2.16	0.073	2.23	0.92	4.46	420	0.55
16p12.1_del	diabetes_insulin_dependent	379089	231	2598	4	1.6	2.50	0.075	2.68	0.89	6.06	424	0.55
15q11.2_dup	diabetes_insulin_dependent	380756	1898	2613	19	13.0	1.46	0.12	1.46	0.90	2.21	604	0.63
15q13.3_del	diabetes_insulin_dependent	378894	36	2595	1	0.2	3.97	0.15	4.42	0.50	16.70	681	0.68
3q29_dup	diabetes_insulin_dependent	378863	5	2594	0	0.03	0.00	0.20	13.53	0.10	121.36	845	0.74
8p23.1_dup	diabetes_insulin_dependent	378863	5	2594	0	0.0	0.00	0.22	12.12	0.09	108.77	890	0.76
15q24_dup	diabetes_insulin_dependent	378866	8	2594	0	0.1	0.00	0.23	10.41	0.08	85.25	944	0.78
NRXN1_del	diabetes_insulin_dependent	379013	155	2596	2	1.1	1.87	0.25	2.28	0.48	6.50	1012	0.79
10q23_dup	diabetes_insulin_dependent	378865	7	2594	0	0.0	0.00	0.28	7.79	0.06	66.90	1108	0.80
13q12dup_CRYL1	diabetes_insulin_dependent	378868	10	2594	0	0.1	0.00	0.28	7.70	0.06	60.93	1109	0.80
17q11.2_del_NF1	diabetes_insulin_dependent	378867	9	2594	0	0.1	0.00	0.30	6.99	0.05	56.49	1167	0.81
3q29_del	diabetes_insulin_dependent	378867	9	2594	0	0.1	0.00	0.32	6.49	0.05	51.98	1215	0.82
15q11q13dup_BP3_BP5	diabetes_insulin_dependent	378867	9	2594	0	0.1	0.00	0.32	6.42	0.05	52.48	1228	0.82
13q12.12_dup	diabetes_insulin_dependent	379070	212	2594	0	1.5	0.00	0.35	0.34	0.00	2.31	1325	0.84
16p12.1_dup	diabetes_insulin_dependent	379048	190	2596	2	1.3	1.53	0.36	1.92	0.40	5.45	1329	0.84
15q11q13del_BP3-BP4	diabetes_insulin_dependent	378872	14	2594	0	0.1	0.00	0.37	5.06	0.04	38.52	1367	0.85
22q11.2_del	diabetes_insulin_dependent	378868	10	2594	0	0.1	0.00	0.38	4.88	0.04	39.19	1406	0.85
22q11.2distal_dup	diabetes_insulin_dependent	378870	12	2594	0	0.1	0.00	0.39	4.65	0.04	35.76	1441	0.85
PWS_dup	diabetes_insulin_dependent	378874	16	2594	0	0.1	0.00	0.39	4.59	0.04	34.45	1454	0.85
WBS_dup	diabetes_insulin_dependent	378871	13	2594	0	0.1	0.00	0.40	4.51	0.04	34.45	1467	0.85
1q21.1_del	diabetes_insulin_dependent	378963	105	2595	1	0.7	1.39	0.45	1.99	0.23	7.22	1617	0.88
17p12_HNPP_del	diabetes_insulin_dependent	379075	217	2596	2	1.5	1.34	0.46	1.67	0.35	4.73	1634	0.88
17q12_dup	diabetes_insulin_dependent	378956	98	2595	1	0.7	1.49	0.48	1.91	0.22	6.94	1699	0.88
7q11.23_dup_distal	diabetes_insulin_dependent	378881	23	2594	0	0.2	0.00	0.48	3.29	0.03	23.83	1719	0.88
15q11.2_del	diabetes_insulin_dependent	380415	1557	2607	13	10.7	1.22	0.51	1.21	0.67	1.98	1781	0.89
16p13.11_del	diabetes_insulin_dependent	378981	123	2595	1	0.8	1.19	0.51	1.80	0.21	6.52	1799	0.89
2q11.2_dup	diabetes_insulin_dependent	378884	26	2594	0	0.2	0.00	0.52	2.95	0.02	21.21	1811	0.90
17p12_CMT1A_dup	diabetes_insulin_dependent	378973	115	2595	1	0.8	1.27	0.56	1.69	0.19	6.12	1910	0.91
16p13.11_dup	diabetes_insulin_dependent	379637	779	2598	4	5.3	0.75	0.62	0.80	0.27	1.79	2069	0.94
16p11.2distal_dup	diabetes_insulin_dependent	378985	127	2594	0	0.9	0.00	0.63	0.54	0.00	3.71	2099	0.94
16p11.2_dup	diabetes_insulin_dependent	378988	130	2595	1	0.9	1.12	0.64	1.50	0.17	5.42	2134	0.94
2q11.2_del	diabetes_insulin_dependent	378887	29	2594	0	0.2	0.00	0.65	2.07	0.02	14.90	2147	0.95
15q13.3_dup	diabetes_insulin_dependent	379080	222	2596	2	1.5	1.31	0.65	1.35	0.28	3.82	2163	0.95
10q11.21q11.23_dup	diabetes_insulin_dependent	378898	40	2594	0	0.3	0.00	0.72	1.74	0.01	12.31	2346	0.96
2q21.1_del	diabetes_insulin_dependent	378898	40	2594	0	0.3	0.00	0.76	1.59	0.01	11.20	2477	0.96
22q11.2_dup	diabetes_insulin_dependent	379122	264	2596	2	1.8	1.11	0.80	1.18	0.25	3.34	2587	0.97
15q11q13dup_BP3_BP4	diabetes_insulin_dependent	378908	50	2594	0	0.3	0.00	0.85	1.32	0.01	9.21	2744	0.97
2q13del(NPHP1)	diabetes_insulin_dependent	381172	2314	2610	16	15.8	1.01	0.86	1.04	0.62	1.64	2771	0.98
2q13dup(NPHP1)	diabetes_insulin_dependent	380713	1855	2607	13	12.7	1.02	0.86	1.05	0.58	1.72	2775	0.98
15q13.3dup(CHRNA7)	diabetes_insulin_dependent	381683	2825	2612	18	19.3	0.93	0.89	0.97	0.59	1.48	2838	0.98
2q13_del	diabetes_insulin_dependent	378909	51	2594	0	0.3	0.00	0.89	1.21	0.01	8.50	2861	0.98
2q21.1_dup	diabetes_insulin_dependent	378913	55	2594	0	0.4	0.00	0.91	1.18	0.01	8.22	2902	0.98
13q12.12_del	diabetes_insulin_dependent	378942	84	2594	0	0.6	0.00	0.92	0.87	0.01	5.97	2920	0.98
13q12del_CRYL1	diabetes_insulin_dependent	379219	361	2596	2	2.5	0.81	0.94	1.05	0.22	2.96	2981	0.99
10q11.21q11.23_del	diabetes_insulin_dependent	378914	56	2594	0	0.4	0.00	0.94	1.11	0.01	7.73	2994	0.99
TAR_del	diabetes_insulin_dependent	378927	69	2594	0	0.5	0.00	0.97	1.06	0.01	7.37	3056	0.99
2q13_dup	diabetes_insulin_dependent	378929	71	2594	0	0.5	0.00	0.99	0.98	0.01	6.80	3105	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2_del	diabetes_other	360794	76	20761	27	4.4	4.82	2.54E-11	6.07	3.77	9.50	2	3.98E-08
16p11.2distal_del	diabetes_other	360757	39	20749	15	2.2	5.11	8.86E-08	6.95	3.67	12.57	6	4.63E-05
1q21.1_dup	diabetes_other	360864	146	20756	22	8.4	2.41	0.00017	2.65	1.64	4.10	21	0.025
16p12.1_del	diabetes_other	360926	208	20761	27	12.0	2.11	0.00021	2.34	1.53	3.46	24	0.028
15q13.3_del	diabetes_other	360746	28	20743	9	1.6	4.48	0.00038	4.87	2.15	10.15	31	0.039
2q13_dup	diabetes_other	360778	60	20745	11	3.5	2.85	0.0012	3.39	1.68	6.29	41	0.094
16p13.11_dup	diabetes_other	361437	719	20798	64	41.4	1.50	0.0018	1.55	1.18	1.99	52	0.11
17p12_HNPP_del	diabetes_other	360915	197	20756	22	11.3	1.85	0.0061	1.97	1.23	3.00	87	0.22
10q11.21q11.23_dup	diabetes_other	360752	34	20740	6	2.0	2.76	0.0073	3.93	1.51	8.76	101	0.23
15q11q13del_BP3-BP4	diabetes_other	360729	11	20737	3	0.6	3.94	0.0092	6.94	1.74	21.45	116	0.25
3q29_del	diabetes_other	360725	7	20736	2	0.4	4.09	0.049	5.57	1.01	21.91	326	0.47
16p11.2distal_dup	diabetes_other	360843	125	20736	2	7.2	0.29	0.051	0.35	0.07	1.00	332	0.48
22q11.2_del	diabetes_other	360726	8	20736	2	0.5	3.68	0.063	5.03	0.90	19.98	374	0.53
TAR_dup	diabetes_other	361095	377	20765	31	21.7	1.40	0.068	1.44	0.97	2.04	389	0.55
3q29_dup	diabetes_other	360722	4	20735	1	0.23	3.68	0.069	8.24	0.81	46.69	400	0.54
22q11.2distal_del	diabetes_other	360722	4	20735	1	0.2	3.68	0.12	5.94	0.56	35.70	597	0.63
10q11.21q11.23_del	diabetes_other	360768	50	20740	6	2.9	1.97	0.12	2.04	0.80	4.42	617	0.63
NRXN1_del	diabetes_other	360863	145	20746	12	8.3	1.41	0.18	1.54	0.81	2.65	766	0.72
17q12_dup	diabetes_other	360808	90	20743	9	5.2	1.67	0.20	1.61	0.76	3.02	840	0.74
22q11.2_dup	diabetes_other	360964	246	20754	20	14.1	1.38	0.21	1.35	0.83	2.09	887	0.76
15q11.2_del	diabetes_other	362188	1470	20834	100	84.6	1.17	0.22	1.14	0.92	1.39	912	0.77
2q21.1_dup	diabetes_other	360772	54	20735	1	3.1	0.33	0.25	0.43	0.05	1.60	984	0.78
15q11q13dup_BP3_BP5	diabetes_other	360726	8	20735	1	0.5	2.04	0.25	3.32	0.34	15.84	997	0.78
7q11.23_dup_distal	diabetes_other	360739	21	20736	2	1.2	1.60	0.29	2.23	0.44	7.13	1117	0.80
2q11.2_dup	diabetes_other	360742	24	20736	2	1.4	1.42	0.32	2.09	0.42	6.56	1227	0.81
17q12_del	diabetes_other	360724	6	20735	1	0.3	2.63	0.32	2.78	0.28	14.39	1234	0.82
15q11q13dup_BP3_BP4	diabetes_other	360767	49	20735	1	2.8	0.37	0.38	0.52	0.06	1.94	1412	0.85
15q13.3dup(CHRNA7)	diabetes_other	363398	2680	20897	163	154.1	1.05	0.39	1.07	0.91	1.26	1432	0.85
15q11.2_dup	diabetes_other	362521	1803	20848	114	103.7	1.09	0.42	1.08	0.89	1.31	1514	0.87
16p13.11_del	diabetes_other	360834	116	20742	8	6.7	1.19	0.44	1.34	0.61	2.57	1564	0.87
2q21.1_del	diabetes_other	360757	39	20735	1	2.2	0.46	0.46	0.57	0.06	2.16	1648	0.88
17p12_CMT1A_dup	diabetes_other	360826	108	20742	8	6.2	1.27	0.51	1.28	0.58	2.46	1796	0.89
1q21.1_del	diabetes_other	360820	102	20738	4	5.9	0.69	0.59	0.78	0.26	1.80	1997	0.93
22q11.2distal_dup	diabetes_other	360730	12	20734	0	0.7	0.00	0.63	0.53	0.00	4.16	2108	0.94
WBS_dup	diabetes_other	360731	13	20734	0	0.7	0.00	0.68	0.58	0.00	4.49	2239	0.95
13q12del_CRYL1	diabetes_other	361065	347	20750	16	19.9	0.81	0.69	0.90	0.53	1.44	2262	0.95
15q24_dup	diabetes_other	360726	8	20734	0	0.5	0.00	0.71	1.79	0.01	14.63	2317	0.96
15q13.3_dup	diabetes_other	360927	209	20749	15	12.0	1.23	0.72	1.10	0.62	1.80	2368	0.96
2q13del(NPHP1)	diabetes_other	362925	2207	20857	123	126.8	0.97	0.74	0.97	0.80	1.16	2397	0.96
15q13.3del(CHRNA7)	diabetes_other	360728	10	20734	0	0.6	0.00	0.75	0.65	0.00	5.29	2436	0.97
2q13_del	diabetes_other	360767	49	20736	2	2.8	0.72	0.79	0.84	0.17	2.53	2564	0.97
17q11.2_del_NF1	diabetes_other	360727	9	20734	0	0.5	0.00	0.80	0.70	0.01	5.75	2571	0.97
Potocki_Lupski	diabetes_other	360723	5	20734	0	0.3	0.00	0.80	1.50	0.01	14.11	2580	0.97
16p11.2_dup	diabetes_other	360841	123	20742	8	7.1	1.12	0.80	1.10	0.50	2.09	2594	0.97
PWS_dup	diabetes_other	360734	16	20734	0	0.9	0.00	0.80	0.71	0.01	5.52	2596	0.97
2q13dup(NPHP1)	diabetes_other	362481	1763	20839	105	101.4	1.03	0.82	1.02	0.83	1.24	2628	0.97
13q12.12_dup	diabetes_other	360920	202	20744	10	11.6	0.87	0.82	0.93	0.47	1.65	2639	0.97
2q11.2_del	diabetes_other	360746	28	20735	1	1.6	0.63	0.83	0.84	0.09	3.28	2677	0.97
8p23.1_dup	diabetes_other	360723	5	20734	0	0.3	0.00	0.84	1.36	0.01	12.41	2714	0.97
13q12.12_del	diabetes_other	360798	80	20738	4	4.6	0.88	0.89	0.93	0.31	2.18	2835	0.98
TAR_del	diabetes_other	360784	66	20737	3	3.8	0.80	0.89	0.93	0.25	2.40	2858	0.98
16p12.1_dup	diabetes_other	360900	182	20744	10	10.5	0.96	0.97	1.01	0.51	1.81	3075	0.99
13q12dup_CRYL1	diabetes_other	360728	10	20734	0	0.6	0.00	0.98	0.97	0.01	7.86	3092	1.00
10q23_dup	diabetes_other	360725	7	20734	0	0.4	0.00	0.99	0.98	0.01	8.93	3104	1.00



CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p13.11_dup	died_to_2016	371226	743	11009	40	22.0	1.78	0.00097	1.79	1.28	2.43	38	0.080
3q29_dup	died_to_2016	370486	3	10971	2	0.09	13.91	0.0013	27.81	4.54	146.05	44	0.093
15q13.3_dup	died_to_2016	370690	207	10986	17	6.1	2.64	0.0017	2.42	1.43	3.85	50	0.11
16p11.2_del	died_to_2016	370576	93	10979	10	2.8	3.38	0.0050	2.88	1.42	5.27	79	0.20
16p12.1_del	died_to_2016	370703	220	10984	15	6.5	2.22	0.0052	2.29	1.31	3.72	80	0.20
13q12.12_del	died_to_2016	370560	77	10976	7	2.3	2.90	0.0092	3.21	1.38	6.41	117	0.25
7q11.23_dup_distal	died_to_2016	370503	20	10972	3	0.6	4.54	0.010	6.34	1.67	17.81	126	0.26
2q21.1_dup	died_to_2016	370533	50	10974	5	1.5	3.16	0.021	3.37	1.23	7.51	193	0.35
1q21.1_del	died_to_2016	370582	99	10976	7	2.9	2.30	0.031	2.53	1.10	5.00	250	0.39
1q21.1_dup	died_to_2016	370641	158	10979	10	4.7	2.07	0.038	2.09	1.05	3.73	278	0.43
17q12_dup	died_to_2016	370575	92	10976	7	2.7	2.46	0.040	2.42	1.04	4.78	295	0.43
Potocki_Lupski	died_to_2016	370487	4	10970	1	0.1	6.96	0.048	10.52	1.02	60.19	322	0.47
13q12.12_dup	died_to_2016	370684	201	10980	11	6.0	1.80	0.051	1.92	1.00	3.33	334	0.48
22q11.2distal_del	died_to_2016	370487	4	10970	1	0.1	6.96	0.052	9.90	0.97	55.97	337	0.49
22q11.2_dup	died_to_2016	370735	252	10983	14	7.5	1.83	0.053	1.77	0.99	2.90	345	0.48
15q24_dup	died_to_2016	370490	7	10970	1	0.2	4.35	0.061	8.45	0.88	39.62	367	0.52
17p12_HNPP_del	died_to_2016	370700	217	10971	2	6.4	0.32	0.071	0.38	0.08	1.07	412	0.54
10q11.21q11.23_dup	died_to_2016	370520	37	10972	3	1.1	2.61	0.075	3.21	0.87	8.49	426	0.55
17q12_del	died_to_2016	370489	6	10970	1	0.2	4.97	0.11	5.90	0.61	28.99	544	0.62
13q12dup_CRYL1	died_to_2016	370492	9	10970	1	0.3	3.48	0.11	5.68	0.61	24.93	548	0.62
15q11q13dup_BP3_BP5	died_to_2016	370491	8	10970	1	0.2	3.86	0.11	5.75	0.60	26.42	551	0.62
15q13.3_del	died_to_2016	370517	34	10972	3	1.0	2.82	0.13	2.63	0.71	7.02	646	0.64
22q11.2_del	died_to_2016	370492	9	10970	1	0.3	3.48	0.17	4.21	0.44	18.98	752	0.72
16p11.2distal_dup	died_to_2016	370604	121	10975	6	3.6	1.64	0.18	1.80	0.73	3.67	784	0.73
PWS_dup	died_to_2016	370498	15	10970	1	0.4	2.17	0.19	3.87	0.42	15.85	801	0.73
16p12.1_dup	died_to_2016	370667	184	10977	8	5.4	1.45	0.22	1.59	0.73	2.98	897	0.76
16p11.2distal_del	died_to_2016	370534	51	10972	3	1.5	1.93	0.24	2.06	0.57	5.32	956	0.78
15q11q13dup_BP3_BP4	died_to_2016	370533	50	10969	0	1.5	0.00	0.30	0.31	0.00	2.14	1175	0.81
TAR_del	died_to_2016	370549	66	10972	3	2.0	1.51	0.31	1.84	0.51	4.71	1196	0.82
15q11.2_dup	died_to_2016	372338	1855	11031	62	55.0	1.12	0.38	1.12	0.86	1.43	1397	0.85
13q12del_CRYL1	died_to_2016	370834	351	10981	12	10.4	1.15	0.39	1.29	0.70	2.18	1450	0.85
2q11.2_dup	died_to_2016	370508	25	10970	1	0.7	1.34	0.43	2.10	0.23	8.14	1538	0.87
15q11.2_del	died_to_2016	372002	1519	11020	51	45.0	1.13	0.47	1.11	0.83	1.45	1668	0.88
2q13dup(NPHP1)	died_to_2016	372303	1820	11017	48	53.9	0.89	0.49	0.90	0.67	1.19	1722	0.89
8p23.1_dup	died_to_2016	370488	5	10969	0	0.1	0.00	0.53	2.94	0.02	26.43	1836	0.90
2q11.2_del	died_to_2016	370512	29	10969	0	0.9	0.00	0.55	0.47	0.00	3.40	1892	0.92
16p13.11_del	died_to_2016	370603	120	10973	4	3.6	1.12	0.57	1.33	0.44	3.04	1945	0.92
17p12_CMT1A_dup	died_to_2016	370595	112	10973	4	3.3	1.20	0.63	1.28	0.42	2.94	2079	0.94
10q11.21q11.23_del	died_to_2016	370537	54	10971	2	1.6	1.24	0.63	1.40	0.29	4.11	2081	0.94
10q23_dup	died_to_2016	370490	7	10969	0	0.2	0.00	0.65	2.08	0.02	17.72	2158	0.95
NRXN1_del	died_to_2016	370635	152	10974	5	4.5	1.11	0.67	1.21	0.45	2.59	2192	0.95
2q13del(NPHP1)	died_to_2016	372750	2267	11032	63	67.1	0.94	0.69	0.95	0.73	1.21	2263	0.95
16p11.2_dup	died_to_2016	370611	128	10972	3	3.8	0.80	0.71	0.82	0.23	2.06	2311	0.96
17q11.2_del_NF1	died_to_2016	370492	9	10969	0	0.3	0.00	0.77	1.59	0.01	12.66	2486	0.97
3q29_del	died_to_2016	370492	9	10969	0	0.3	0.00	0.78	1.53	0.01	12.28	2536	0.97
2q13_dup	died_to_2016	370552	69	10971	2	2.0	0.98	0.81	1.17	0.24	3.40	2624	0.97
2q21.1_del	died_to_2016	370522	39	10970	1	1.2	0.87	0.85	1.17	0.13	4.40	2749	0.97
TAR_dup	died_to_2016	370880	397	10980	11	11.8	0.94	0.86	0.95	0.50	1.63	2768	0.97
15q11q13del_BP3-BP4	died_to_2016	370497	14	10969	0	0.4	0.00	0.87	1.29	0.01	9.76	2778	0.98
2q13_del	died_to_2016	370533	50	10970	1	1.5	0.68	0.89	0.89	0.10	3.32	2841	0.98
15q13.3del(CHRNA7)	died_to_2016	370493	10	10969	0	0.3	0.00	0.92	1.17	0.01	9.32	2922	0.98
WBS_dup	died_to_2016	370496	13	10969	0	0.4	0.00	0.92	1.16	0.01	8.89	2925	0.98
15q13.3dup(CHRNA7)	died_to_2016	373245	2762	11050	81	81.8	0.99	0.92	1.01	0.80	1.25	2938	0.99
22q11.2distal_dup	died_to_2016	370495	12	10969	0	0.4	0.00	0.94	1.11	0.01	8.61	2987	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
17q12_del	digestive	299609	1	81850	6	0.3	3.99	0.00044	15.44	3.17	150.30	33	0.042*
17p12_CMT1A_dup	digestive	299685	77	81883	39	21.0	1.57	0.0017	1.91	1.29	2.81	48	0.11
7q11.23_dup_distal	digestive	299631	23	81844	0	6.3	0.00	0.0064	0.08	0.00	0.60	89	0.23
NRXN1_del	digestive	299718	110	81891	47	30.1	1.40	0.0065	1.64	1.15	2.30	91	0.22
22q11.2_dup	digestive	299799	191	81919	75	52.2	1.31	0.012	1.43	1.09	1.87	135	0.27
TAR_dup	digestive	299909	301	81951	107	82.2	1.22	0.013	1.34	1.07	1.67	139	0.28
16p13.11_del	digestive	299696	88	81880	36	24.0	1.35	0.035	1.55	1.03	2.26	262	0.41
17q11.2_del_NF1	digestive	299612	4	81849	5	1.1	2.59	0.037	3.87	1.09	14.46	273	0.42
8p23.1_dup	digestive	299610	2	81847	3	0.5	2.80	0.056	4.92	0.95	29.63	356	0.50
16p13.11_dup	digestive	300242	634	81993	149	173.1	0.89	0.094	0.86	0.72	1.03	505	0.58
16p11.2_del	digestive	299681	73	81874	30	19.9	1.36	0.099	1.45	0.93	2.20	526	0.59
16p12.1_del	digestive	299782	174	81905	61	47.5	1.21	0.10	1.28	0.95	1.71	536	0.61
10q23_dup	digestive	299612	4	81847	3	1.1	2.00	0.11	3.38	0.73	14.49	564	0.63
13q12del_CRYL1	digestive	299886	278	81929	85	75.9	1.09	0.11	1.22	0.95	1.55	568	0.63
17q12_dup	digestive	299679	71	81872	28	19.4	1.32	0.12	1.44	0.91	2.21	578	0.63
1q21.1_dup	digestive	299731	123	81889	45	33.6	1.25	0.12	1.32	0.93	1.85	589	0.63
3q29_dup	digestive	299611	3	81846	2	0.82	1.86	0.16	3.56	0.58	18.94	710	0.69
2q13_dup	digestive	299669	61	81854	10	16.7	0.66	0.16	0.64	0.31	1.18	722	0.69
13q12dup_CRYL1	digestive	299614	6	81848	4	1.6	1.86	0.17	2.41	0.66	8.03	749	0.71
1q21.1_del	digestive	299687	79	81871	27	21.6	1.19	0.26	1.29	0.82	1.98	1032	0.79
16p11.2_dup	digestive	299705	97	81878	34	26.5	1.21	0.27	1.26	0.84	1.84	1043	0.80
WBS_dup	digestive	299620	12	81845	1	3.3	0.36	0.27	0.42	0.05	1.77	1058	0.80
Potocki_Lupski	digestive	299611	3	81846	2	0.8	1.86	0.29	2.49	0.41	12.88	1135	0.81
15q11q13dup_BP3_BP5	digestive	299614	6	81847	3	1.6	1.55	0.30	2.08	0.48	7.55	1158	0.81
13q12.12_dup	digestive	299781	173	81883	39	47.3	0.86	0.31	0.84	0.58	1.17	1203	0.82
15q11q13del_BP3-BP4	digestive	299618	10	81848	4	2.7	1.33	0.33	1.78	0.52	5.14	1249	0.82
15q13.3_del	digestive	299634	26	81855	11	7.1	1.39	0.36	1.40	0.66	2.76	1346	0.85
22q11.2distal_del	digestive	299613	5	81844	0	1.4	0.00	0.37	0.32	0.00	2.90	1366	0.85
15q13.3dup(CHRNA7)	digestive	301868	2260	82427	583	617.1	0.96	0.37	0.96	0.87	1.05	1377	0.85
PWS_dup	digestive	299620	12	81848	4	3.3	1.17	0.38	1.67	0.50	4.68	1393	0.85
2q13_del	digestive	299651	43	81852	8	11.7	0.73	0.40	0.73	0.33	1.46	1468	0.85
2q11.2_del	digestive	299629	21	81852	8	5.7	1.29	0.40	1.43	0.60	3.09	1471	0.85
15q11q13dup_BP3_BP4	digestive	299650	42	81852	8	11.5	0.75	0.42	0.74	0.33	1.49	1516	0.87
16p12.1_dup	digestive	299764	156	81880	36	42.6	0.87	0.43	0.87	0.59	1.23	1549	0.87
15q13.3_dup	digestive	299787	179	81889	45	48.9	0.94	0.50	0.89	0.64	1.23	1754	0.89
15q24_dup	digestive	299614	6	81846	2	1.6	1.17	0.50	1.70	0.31	6.76	1756	0.89
10q11.21q11.23_dup	digestive	299642	34	81850	6	9.3	0.70	0.56	0.78	0.30	1.70	1900	0.92
3q29_del	digestive	299616	8	81845	1	2.2	0.52	0.56	0.61	0.06	2.75	1922	0.92
2q21.1_del	digestive	299638	30	81854	10	8.2	1.17	0.59	1.22	0.57	2.40	1988	0.93
16p11.2distal_dup	digestive	299710	102	81869	25	27.9	0.92	0.65	0.90	0.57	1.38	2141	0.95
10q11.21q11.23_del	digestive	299653	45	81855	11	12.3	0.92	0.67	0.87	0.43	1.61	2203	0.95
2q13dup(NPHP1)	digestive	301070	1462	82250	406	399.4	1.01	0.70	1.02	0.91	1.14	2288	0.96
15q11.2_dup	digestive	301105	1497	82264	420	409.0	1.02	0.70	1.02	0.91	1.14	2289	0.95
15q11.2_del	digestive	300847	1239	82175	331	338.4	0.98	0.71	0.98	0.86	1.10	2309	0.96
TAR_del	digestive	299664	56	81857	13	15.3	0.88	0.78	0.92	0.48	1.62	2517	0.97
17p12_HNPP_del	digestive	299778	170	81893	49	46.4	1.04	0.81	1.04	0.75	1.42	2622	0.97
22q11.2_del	digestive	299616	8	81846	2	2.2	0.93	0.84	1.16	0.22	4.31	2693	0.97
22q11.2distal_dup	digestive	299617	9	81847	3	2.5	1.17	0.84	1.14	0.28	3.64	2696	0.97
16p11.2distal_del	digestive	299650	42	81856	12	11.5	1.04	0.84	1.07	0.54	1.95	2721	0.97
2q21.1_dup	digestive	299651	43	81856	12	11.7	1.02	0.89	1.05	0.53	1.92	2831	0.98
13q12.12_del	digestive	299674	66	81862	18	18.0	1.00	0.91	0.97	0.56	1.60	2905	0.98
15q13.3del(CHRNA7)	digestive	299616	8	81846	2	2.2	0.93	0.96	1.04	0.19	3.82	3033	0.99
2q11.2_dup	digestive	299629	21	81849	5	5.7	0.90	0.97	0.98	0.34	2.37	3065	0.99
2q13del(NPHP1)	digestive	301439	1831	82343	499	500.2	1.00	0.97	1.00	0.90	1.10	3073	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
3q29_dup	diverticular_disease_intestine	354321	2	27136	3	0.15	8.44	0.00010	41.76	7.38	276.02	19	0.017*
17q11.2_del_NF1	diverticular_disease_intestine	354325	6	27136	3	0.5	4.69	0.022	5.66	1.32	20.54	203	0.34
2q13_del	diverticular_disease_intestine	354363	44	27140	7	3.4	1.93	0.026	2.73	1.14	5.71	224	0.37
15q11.2_del	diverticular_disease_intestine	355797	1478	27225	92	113.1	0.82	0.059	0.82	0.66	1.01	361	0.51
16p13.11_dup	diverticular_disease_intestine	355035	716	27200	67	54.9	1.20	0.065	1.28	0.98	1.64	378	0.54
16p11.2_del	diverticular_disease_intestine	354420	101	27135	2	7.7	0.27	0.071	0.37	0.08	1.08	414	0.54
15q11.2_dup	diverticular_disease_intestine	356120	1801	27249	116	137.8	0.85	0.086	0.85	0.70	1.02	473	0.57
16p11.2distal_dup	diverticular_disease_intestine	354434	115	27145	12	8.8	1.33	0.13	1.64	0.86	2.86	622	0.64
17q12_dup	diverticular_disease_intestine	354415	96	27136	3	7.4	0.43	0.13	0.48	0.13	1.21	642	0.64
16p12.1_del	diverticular_disease_intestine	354532	213	27155	22	16.3	1.32	0.14	1.42	0.89	2.16	658	0.65
15q13.3_dup	diverticular_disease_intestine	354533	214	27143	10	16.4	0.63	0.17	0.66	0.33	1.17	740	0.70
16p11.2_dup	diverticular_disease_intestine	354437	118	27146	13	9.0	1.40	0.17	1.52	0.82	2.61	757	0.72
2q21.1_dup	diverticular_disease_intestine	354368	49	27139	6	3.8	1.53	0.18	1.84	0.73	3.95	777	0.72
15q13.3dup(CHRNA7)	diverticular_disease_intestine	356982	2663	27313	180	203.7	0.89	0.19	0.90	0.77	1.05	804	0.73
TAR_dup	diverticular_disease_intestine	354693	374	27167	34	28.6	1.17	0.19	1.28	0.88	1.79	805	0.73
15q24_dup	diverticular_disease_intestine	354326	7	27134	1	0.5	1.76	0.19	4.07	0.41	20.22	806	0.73
10q23_dup	diverticular_disease_intestine	354325	6	27134	1	0.5	2.01	0.19	4.10	0.40	21.91	816	0.74
2q11.2_del	diverticular_disease_intestine	354348	29	27133	0	2.2	0.00	0.21	0.24	0.00	1.77	876	0.75
NRXN1_del	diverticular_disease_intestine	354462	143	27147	14	11.0	1.25	0.22	1.43	0.79	2.40	899	0.76
2q11.2_dup	diverticular_disease_intestine	354342	23	27136	3	1.8	1.62	0.24	2.12	0.56	5.98	964	0.78
22q11.2_del	diverticular_disease_intestine	354328	9	27134	1	0.7	1.41	0.25	3.29	0.34	15.12	991	0.78
16p11.2distal_del	diverticular_disease_intestine	354368	49	27138	5	3.8	1.30	0.28	1.70	0.62	3.80	1084	0.80
13q12.12_dup	diverticular_disease_intestine	354513	194	27151	18	14.9	1.19	0.30	1.30	0.78	2.06	1153	0.81
7q11.23_dup_distal	diverticular_disease_intestine	354342	23	27133	0	1.8	0.00	0.30	0.30	0.00	2.19	1164	0.81
2q13del(NPHP1)	diverticular_disease_intestine	356495	2176	27287	154	166.6	0.93	0.30	0.92	0.77	1.08	1168	0.81
17p12_HNPP_del	diverticular_disease_intestine	354518	199	27153	20	15.2	1.28	0.32	1.27	0.78	1.97	1220	0.82
16p13.11_del	diverticular_disease_intestine	354432	113	27144	11	8.7	1.25	0.33	1.37	0.70	2.44	1262	0.82
3q29_del	diverticular_disease_intestine	354327	8	27134	1	0.6	1.56	0.34	2.59	0.27	11.92	1295	0.83
15q13.3_del	diverticular_disease_intestine	354355	36	27134	1	2.8	0.38	0.39	0.52	0.06	1.97	1426	0.85
13q12.12_del	diverticular_disease_intestine	354395	76	27141	8	5.8	1.34	0.44	1.34	0.60	2.60	1590	0.87
PWS_dup	diverticular_disease_intestine	354334	15	27134	1	1.1	0.88	0.46	2.05	0.22	8.90	1638	0.88
13q12dup_CRYL1	diverticular_disease_intestine	354328	9	27134	1	0.7	1.41	0.47	2.02	0.21	9.08	1661	0.88
15q11q13del_BP3-BP4	diverticular_disease_intestine	354332	13	27134	1	1.0	1.00	0.50	1.90	0.21	8.00	1747	0.89
2q13_dup	diverticular_disease_intestine	354387	68	27136	3	5.2	0.59	0.51	0.71	0.19	1.81	1784	0.89
22q11.2distal_dup	diverticular_disease_intestine	354331	12	27133	0	0.9	0.00	0.51	0.43	0.00	3.37	1789	0.89
15q11q13dup_BP3_BP4	diverticular_disease_intestine	354365	46	27137	4	3.5	1.12	0.52	1.40	0.45	3.40	1814	0.90
TAR_del	diverticular_disease_intestine	354385	66	27136	3	5.1	0.61	0.56	0.73	0.20	1.89	1917	0.91
10q11.21q11.23_dup	diverticular_disease_intestine	354358	39	27134	1	3.0	0.35	0.60	0.66	0.07	2.53	2019	0.94
WBS_dup	diverticular_disease_intestine	354332	13	27133	0	1.0	0.00	0.63	0.53	0.00	4.15	2100	0.94
10q11.21q11.23_del	diverticular_disease_intestine	354372	53	27136	3	4.1	0.75	0.70	0.81	0.22	2.11	2287	0.96
2q13dup(NPHP1)	diverticular_disease_intestine	356051	1732	27269	136	132.6	1.02	0.72	1.03	0.86	1.23	2352	0.96
2q21.1_del	diverticular_disease_intestine	354356	37	27136	3	2.8	1.05	0.75	1.20	0.32	3.19	2443	0.97
22q11.2distal_del	diverticular_disease_intestine	354324	5	27133	0	0.4	0.00	0.77	1.59	0.01	15.46	2503	0.97
1q21.1_dup	diverticular_disease_intestine	354475	156	27145	12	11.9	1.00	0.80	1.08	0.57	1.86	2572	0.97
15q13.3del(CHRNA7)	diverticular_disease_intestine	354329	10	27133	0	0.8	0.00	0.83	0.74	0.01	5.99	2674	0.97
15q11q13dup_BP3_BP5	diverticular_disease_intestine	354328	9	27133	0	0.7	0.00	0.84	0.75	0.01	6.54	2708	0.97
17p12_CMT1A_dup	diverticular_disease_intestine	354427	108	27141	8	8.3	0.97	0.85	1.07	0.49	2.05	2727	0.97
22q11.2_dup	diverticular_disease_intestine	354568	249	27150	17	19.1	0.90	0.85	0.95	0.55	1.53	2741	0.97
1q21.1_del	diverticular_disease_intestine	354418	99	27140	7	7.6	0.93	0.87	1.07	0.46	2.12	2782	0.98
13q12del_CRYL1	diverticular_disease_intestine	354660	341	27155	22	26.1	0.85	0.89	0.97	0.61	1.46	2848	0.98
Potocki_Lupski	diverticular_disease_intestine	354324	5	27133	0	0.4	0.00	0.91	1.20	0.01	10.92	2888	0.98
17q12_del	diverticular_disease_intestine	354326	7	27133	0	0.5	0.00	0.96	0.93	0.01	8.04	3047	0.99
16p12.1_dup	diverticular_disease_intestine	354498	179	27146	13	13.7	0.95	0.97	1.01	0.55	1.70	3060	0.99
8p23.1_dup	diverticular_disease_intestine	354324	5	27133	0	0.4	0.00	0.97	1.05	0.01	9.59	3072	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q13del(NPHP1)	ear	369500	2267	14282	63	87.6	0.73	0.0069	0.72	0.56	0.92	92	0.23
3q29_dup	ear	367237	4	14220	1	0.15	5.37	0.044	11.03	1.09	61.03	305	0.45
22q11.2distal_del	ear	367237	4	14220	1	0.2	5.37	0.061	8.92	0.88	49.29	366	0.52
Potocki_Lupski	ear	367237	4	14220	1	0.2	5.37	0.065	8.41	0.84	45.59	380	0.54
8p23.1_dup	ear	367237	4	14220	1	0.2	5.37	0.069	8.14	0.81	44.20	395	0.55
16p12.1_del	ear	367454	221	14233	14	8.6	1.60	0.075	1.69	0.95	2.77	423	0.55
15q24_dup	ear	367240	7	14220	1	0.3	3.35	0.096	6.27	0.66	29.15	510	0.59
22q11.2_dup	ear	367484	251	14234	15	9.7	1.51	0.097	1.59	0.91	2.57	518	0.59
17p12_HNPP_del	ear	367439	206	14232	13	8.0	1.59	0.098	1.65	0.90	2.75	523	0.59
10q23_dup	ear	367239	6	14220	1	0.2	3.83	0.10	6.19	0.64	30.31	527	0.60
3q29_del	ear	367241	8	14220	1	0.3	2.98	0.16	4.50	0.48	20.14	704	0.69
22q11.2_del	ear	367242	9	14220	1	0.3	2.68	0.16	4.45	0.47	19.62	714	0.69
15q13.3del(CHRNA7)	ear	367242	9	14220	1	0.3	2.68	0.17	4.17	0.44	18.25	753	0.72
2q11.2_dup	ear	367257	24	14221	2	0.9	2.06	0.17	2.83	0.57	8.74	755	0.72
16p11.2distal_del	ear	367287	54	14219	0	2.1	0.00	0.18	0.23	0.00	1.62	781	0.73
17q12_dup	ear	367326	93	14225	6	3.6	1.63	0.19	1.77	0.72	3.63	822	0.74
16p11.2_del	ear	367330	97	14225	6	3.8	1.56	0.21	1.74	0.70	3.56	878	0.75
2q13dup(NPHP1)	ear	369021	1788	14299	80	69.3	1.15	0.21	1.16	0.92	1.44	883	0.75
2q11.2_del	ear	367260	27	14221	2	1.0	1.85	0.25	2.38	0.48	7.26	982	0.78
16p12.1_dup	ear	367421	188	14223	4	7.3	0.56	0.28	0.62	0.21	1.41	1098	0.80
2q13_dup	ear	367300	67	14223	4	2.6	1.51	0.28	1.76	0.58	4.11	1111	0.80
NRXN1_del	ear	367387	154	14222	3	6.0	0.51	0.28	0.59	0.16	1.46	1114	0.80
22q11.2distal_dup	ear	367244	11	14220	1	0.4	2.24	0.29	2.86	0.31	12.06	1127	0.81
17p12_CMT1A_dup	ear	367343	110	14225	6	4.3	1.39	0.33	1.52	0.62	3.10	1264	0.82
15q13.3_dup	ear	367446	213	14230	11	8.2	1.32	0.34	1.36	0.71	2.35	1282	0.82
2q21.1_dup	ear	367285	52	14222	3	2.0	1.46	0.36	1.72	0.47	4.42	1347	0.85
10q11.21q11.23_dup	ear	367273	40	14219	0	1.5	0.00	0.38	0.35	0.00	2.49	1413	0.85
15q11.2_dup	ear	369072	1839	14297	78	71.2	1.09	0.40	1.10	0.87	1.37	1484	0.85
15q13.3_del	ear	367268	35	14221	2	1.4	1.45	0.47	1.66	0.34	4.96	1684	0.88
13q12.12_del	ear	367313	80	14223	4	3.1	1.28	0.51	1.40	0.46	3.23	1795	0.89
TAR_dup	ear	367624	391	14236	17	15.1	1.12	0.52	1.18	0.70	1.84	1818	0.90
16p13.11_del	ear	16p13.11	121	14222	3	4.7	0.65	0.58	0.75	0.21	1.87	1947	0.93
10q11.21q11.23_del	ear	367288	55	14220	1	2.1	0.48	0.60	0.66	0.08	2.45	2007	0.93
15q13.3dup(CHRNA7)	ear	369968	2735	14327	108	105.9	1.02	0.63	1.05	0.86	1.26	2111	0.94
1q21.1_dup	ear	367394	161	14226	7	6.2	1.12	0.67	1.18	0.51	2.28	2216	0.95
7q11.23_dup_distal	ear	367256	23	14219	0	0.9	0.00	0.67	0.58	0.00	4.16	2220	0.95
2q13_del	ear	367283	50	14220	1	1.9	0.53	0.77	0.79	0.09	2.92	2487	0.97
17q12_del	ear	367240	7	14219	0	0.3	0.00	0.77	1.58	0.01	13.21	2491	0.97
15q11q13dup_BP3_BP4	ear	367282	49	14220	1	1.9	0.54	0.78	0.80	0.09	2.96	2529	0.97
1q21.1_del	ear	367336	103	14222	3	4.0	0.76	0.80	0.87	0.24	2.19	2591	0.97
16p11.2_dup	ear	367360	127	14223	4	4.9	0.82	0.81	0.89	0.30	2.04	2621	0.97
15q11q13dup_BP3_BP5	ear	367242	9	14219	0	0.3	0.00	0.84	1.35	0.01	10.85	2716	0.97
13q12.12_dup	ear	367438	205	14226	7	7.9	0.89	0.86	0.94	0.41	1.81	2774	0.98
13q12dup_CRYL1	ear	367243	10	14219	0	0.4	0.00	0.87	1.28	0.01	9.96	2788	0.98
15q11.2_del	ear	368743	1510	14279	60	58.5	1.03	0.87	1.02	0.78	1.31	2795	0.98
17q11.2_del_NF1	ear	367242	9	14219	0	0.3	0.00	0.90	1.20	0.01	9.52	2882	0.98
13q12del_CRYL1	ear	367584	351	14231	12	13.6	0.89	0.91	0.97	0.52	1.63	2900	0.98
2q21.1_del	ear	367272	39	14220	1	1.5	0.67	0.93	0.93	0.10	3.48	2955	0.99
16p11.2distal_dup	ear	367356	123	14223	4	4.8	0.84	0.94	0.97	0.32	2.21	2992	0.99
16p13.11_dup	ear	367987	754	14248	29	29.2	0.99	0.96	1.01	0.68	1.43	3028	0.99
WBS_dup	ear	367246	13	14219	0	0.5	0.00	0.96	0.93	0.01	7.06	3030	0.99
PWS_dup	ear	367249	16	14219	0	0.6	0.00	0.97	0.95	0.01	7.04	3061	0.99
TAR_del	ear	367300	67	14221	2	2.6	0.78	0.98	0.98	0.20	2.85	3079	0.99
15q11q13del_BP3-BP4	ear	367247	14	14219	0	0.5	0.00	1.00	1.00	0.01	7.50	3130	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q21.1_dup	endocrine_other	379145	53	2362	2	0.3	5.88	0.016	7.85	1.62	23.01	161	0.30
22q11.2_dup	endocrine_other	379353	261	2365	5	1.6	3.04	0.017	3.41	1.28	7.21	171	0.32
16p11.2distal_dup	endocrine_other	379216	124	2363	3	0.8	3.82	0.024	4.56	1.27	11.41	211	0.36
16p13.11_dup	endocrine_other	379866	774	2369	9	4.8	1.86	0.064	1.96	0.96	3.51	375	0.53
1q21.1_del	endocrine_other	379196	104	2362	2	0.6	3.05	0.084	3.84	0.80	11.02	468	0.56
2q13del(NPHP1)	endocrine_other	381401	2309	2381	21	14.4	1.46	0.087	1.49	0.94	2.22	478	0.57
15q13.3_del	endocrine_other	379128	36	2361	1	0.2	4.37	0.088	6.22	0.70	23.42	481	0.57
2q21.1_del	endocrine_other	379131	39	2361	1	0.2	4.04	0.092	6.03	0.68	22.57	495	0.58
17p12_CMT1A_dup	endocrine_other	379206	114	2362	2	0.7	2.79	0.11	3.46	0.72	9.91	538	0.61
2q13_del	endocrine_other	379142	50	2361	1	0.3	3.17	0.12	5.13	0.58	19.01	581	0.63
15q13.3_dup	endocrine_other	379313	221	2363	3	1.4	2.16	0.12	2.62	0.73	6.50	608	0.63
3q29_dup	endocrine_other	379097	5	2360	0	0.03	0.00	0.16	20.43	0.15	188.85	711	0.69
8p23.1_dup	endocrine_other	379097	5	2360	0	0.0	0.00	0.19	14.38	0.11	130.12	817	0.74
22q11.2distal_del	endocrine_other	379097	5	2360	0	0.0	0.00	0.20	13.60	0.10	125.43	846	0.74
2q13_dup	endocrine_other	379162	70	2361	1	0.4	2.28	0.20	3.54	0.40	12.97	854	0.74
Potocki Lupski	endocrine_other	379097	5	2360	0	0.0	0.00	0.20	13.31	0.10	119.54	855	0.74
10q23_dup	endocrine_other	379099	7	2360	0	0.0	0.00	0.21	12.62	0.10	105.94	871	0.75
17q12_del	endocrine_other	379099	7	2360	0	0.0	0.00	0.22	11.52	0.09	95.08	903	0.76
15q24_dup	endocrine_other	379100	8	2360	0	0.0	0.00	0.25	9.30	0.07	75.96	1008	0.78
15q13.3del(CHRNA7)	endocrine_other	379102	10	2360	0	0.1	0.00	0.27	8.51	0.07	66.77	1044	0.80
3q29_del	endocrine_other	379101	9	2360	0	0.1	0.00	0.27	8.26	0.06	66.05	1060	0.80
17q11.2_del_NF1	endocrine_other	379101	9	2360	0	0.1	0.00	0.27	8.20	0.06	65.16	1064	0.80
22q11.2_del	endocrine_other	379102	10	2360	0	0.1	0.00	0.27	8.10	0.06	63.52	1071	0.80
15q11q13dup_BP3_BP5	endocrine_other	379101	9	2360	0	0.1	0.00	0.28	8.02	0.06	64.34	1083	0.80
13q12dup_CRYL1	endocrine_other	379102	10	2360	0	0.1	0.00	0.30	6.96	0.05	54.46	1166	0.81
16p12.1_dup	endocrine_other	379282	190	2362	2	1.2	1.68	0.31	2.07	0.43	5.88	1181	0.81
17q12_dup	endocrine_other	379190	98	2361	1	0.6	1.63	0.31	2.62	0.30	9.50	1198	0.81
15q11q13del_BP3-BP4	endocrine_other	379106	14	2360	0	0.1	0.00	0.33	5.92	0.05	44.57	1267	0.82
WBS_dup	endocrine_other	379105	13	2360	0	0.1	0.00	0.34	5.86	0.05	45.18	1277	0.83
22q11.2distal_dup	endocrine_other	379104	12	2360	0	0.1	0.00	0.35	5.46	0.04	42.24	1322	0.83
PWS_dup	endocrine_other	379108	16	2360	0	0.1	0.00	0.37	5.13	0.04	38.19	1355	0.84
17p12_HNPP_del	endocrine_other	379309	217	2362	2	1.4	1.48	0.38	1.85	0.39	5.25	1399	0.85
13q12.12_dup	endocrine_other	379304	212	2360	0	1.3	0.00	0.42	0.38	0.00	2.58	1504	0.87
7q11.23_dup_distal	endocrine_other	379115	23	2360	0	0.1	0.00	0.46	3.55	0.03	25.66	1645	0.88
TAR_dup	endocrine_other	379499	407	2361	1	2.5	0.40	0.50	0.61	0.07	2.16	1772	0.89
2q11.2_dup	endocrine_other	379118	26	2360	0	0.2	0.00	0.50	3.08	0.02	22.14	1774	0.89
2q11.2_del	endocrine_other	379121	29	2360	0	0.2	0.00	0.53	2.85	0.02	20.33	1840	0.90
15q11.2_del	endocrine_other	380651	1559	2371	11	9.7	1.13	0.56	1.20	0.63	2.04	1912	0.91
NRXN1_del	endocrine_other	379248	156	2361	1	1.0	1.03	0.60	1.58	0.18	5.68	2021	0.94
10q11.21q11.23_dup	endocrine_other	379132	40	2360	0	0.2	0.00	0.61	2.27	0.02	15.99	2045	0.94
15q13.3dup(CHRNA7)	endocrine_other	381916	2824	2379	19	17.6	1.08	0.64	1.11	0.69	1.69	2135	0.94
1q21.1_dup	endocrine_other	379259	167	2361	1	1.0	0.96	0.70	1.39	0.16	5.01	2295	0.96
16p11.2_dup	endocrine_other	379223	131	2360	0	0.8	0.00	0.70	0.61	0.00	4.16	2299	0.96
15q11q13dup_BP3_BP4	endocrine_other	379142	50	2360	0	0.3	0.00	0.71	1.77	0.01	12.38	2316	0.96
16p13.11_del	endocrine_other	379216	124	2360	0	0.8	0.00	0.72	0.62	0.00	4.26	2333	0.96
2q13dup(NPHP1)	endocrine_other	380948	1856	2372	12	11.6	1.04	0.76	1.09	0.59	1.82	2474	0.96
16p11.2distal_del	endocrine_other	379146	54	2360	0	0.3	0.00	0.79	1.51	0.01	10.52	2544	0.97
13q12del_CRYL1	endocrine_other	379453	361	2362	2	2.2	0.89	0.83	1.15	0.24	3.25	2662	0.97
16p11.2_del	endocrine_other	379195	103	2360	0	0.6	0.00	0.84	0.77	0.01	5.26	2722	0.97
10q11.21q11.23_del	endocrine_other	379148	56	2360	0	0.3	0.00	0.86	1.29	0.01	8.95	2777	0.98
15q11.2_dup	endocrine_other	380997	1905	2372	12	11.9	1.01	0.87	1.05	0.57	1.75	2802	0.98
TAR_del	endocrine_other	379161	69	2360	0	0.4	0.00	0.90	1.21	0.01	8.36	2869	0.98
13q12.12_del	endocrine_other	379176	84	2360	0	0.5	0.00	0.95	0.92	0.01	6.32	3018	0.99
16p12.1_del	endocrine_other	379326	234	2361	1	1.5	0.69	0.99	1.01	0.12	3.61	3114	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
22q11.2_dup	gastric_reflux	347374	217	34344	49	21.5	2.05	1.82E-06	2.31	1.67	3.13	8	0.00071
15q11q13del_BP3-BP4	gastric_reflux	347165	8	34301	6	0.8	4.77	0.00018	9.11	3.11	25.43	23	0.025
16p12.1_del	gastric_reflux	347359	202	34328	33	20.0	1.56	0.0098	1.67	1.14	2.38	123	0.25
10q23_dup	gastric_reflux	347162	5	34297	2	0.5	3.18	0.055	5.44	0.96	23.29	353	0.49
17p12_CMT1A_dup	gastric_reflux	347257	100	34311	16	9.9	1.53	0.064	1.69	0.97	2.78	376	0.53
15q11q13dup_BP3_BP4	gastric_reflux	347199	42	34303	8	4.1	1.78	0.068	2.12	0.94	4.23	392	0.54
13q12.12_del	gastric_reflux	347229	72	34307	12	7.1	1.59	0.11	1.70	0.89	3.00	540	0.62
1q21.1_del	gastric_reflux	347249	92	34309	14	9.1	1.47	0.11	1.61	0.88	2.72	573	0.63
15q13.3dup(CHRNA7)	gastric_reflux	349772	2615	34523	228	258.1	0.89	0.14	0.90	0.79	1.03	657	0.65
13q12dup_CRYL1	gastric_reflux	347165	8	34297	2	0.8	2.22	0.18	2.92	0.55	10.63	782	0.73
16p13.11_del	gastric_reflux	347266	109	34310	15	10.8	1.35	0.20	1.45	0.81	2.39	828	0.74
1q21.1_dup	gastric_reflux	347305	148	34315	20	14.6	1.32	0.20	1.37	0.84	2.13	838	0.74
10q11.21q11.23_dup	gastric_reflux	347192	35	34300	5	3.5	1.39	0.24	1.79	0.65	4.08	959	0.78
15q11.2_dup	gastric_reflux	348888	1731	34481	186	171.1	1.08	0.27	1.09	0.93	1.27	1048	0.80
2q11.2_del	gastric_reflux	347182	25	34299	4	2.5	1.53	0.28	1.81	0.57	4.51	1106	0.80
16p11.2_del	gastric_reflux	347248	91	34307	12	9.0	1.30	0.32	1.37	0.72	2.39	1221	0.82
10q11.21q11.23_del	gastric_reflux	347206	49	34302	7	4.8	1.39	0.34	1.48	0.63	3.02	1298	0.83
16p12.1_dup	gastric_reflux	347336	179	34308	13	17.7	0.75	0.34	0.77	0.42	1.29	1300	0.83
17q12_dup	gastric_reflux	347250	93	34301	6	9.2	0.67	0.35	0.70	0.28	1.43	1316	0.83
17p12_HNPP_del	gastric_reflux	347352	195	34319	24	19.3	1.22	0.35	1.23	0.79	1.84	1319	0.83
15q13.3_dup	gastric_reflux	347365	208	34311	16	20.5	0.79	0.36	0.80	0.46	1.27	1334	0.84
2q13del(NPHP1)	gastric_reflux	349290	2133	34492	197	210.6	0.94	0.36	0.93	0.80	1.08	1339	0.84
13q12del_CRYL1	gastric_reflux	347485	328	34330	35	32.4	1.07	0.37	1.18	0.82	1.64	1381	0.85
2q13_dup	gastric_reflux	347220	63	34303	8	6.2	1.25	0.38	1.40	0.63	2.72	1401	0.85
15q24_dup	gastric_reflux	347164	7	34296	1	0.7	1.39	0.39	2.36	0.25	11.04	1424	0.85
22q11.2distal_dup	gastric_reflux	347169	12	34295	0	1.2	0.00	0.40	0.36	0.00	2.73	1476	0.85
WBS_dup	gastric_reflux	347170	13	34295	0	1.3	0.00	0.42	0.37	0.00	2.81	1513	0.87
17q12_del	gastric_reflux	347163	6	34296	1	0.6	1.59	0.43	2.18	0.22	10.53	1556	0.87
PWS_dup	gastric_reflux	347173	16	34295	0	1.6	0.00	0.44	0.38	0.00	2.86	1565	0.87
16p11.2distal_del	gastric_reflux	347208	51	34298	3	5.0	0.62	0.48	0.69	0.19	1.78	1713	0.88
22q11.2_del	gastric_reflux	347166	9	34296	1	0.9	1.11	0.53	1.83	0.19	8.05	1830	0.90
3q29_del	gastric_reflux	347165	8	34296	1	0.8	1.24	0.55	1.77	0.19	7.92	1881	0.91
15q13.3del(CHRNA7)	gastric_reflux	347167	10	34295	0	1.0	0.00	0.59	0.49	0.00	3.87	1979	0.93
2q13dup(NPHP1)	gastric_reflux	348852	1695	34468	173	167.5	1.03	0.60	1.04	0.89	1.22	2017	0.94
16p11.2_dup	gastric_reflux	347278	121	34305	10	12.0	0.85	0.62	0.85	0.43	1.53	2059	0.94
16p13.11_dup	gastric_reflux	347874	717	34361	66	70.8	0.94	0.62	0.94	0.72	1.20	2071	0.94
15q11q13dup_BP3_BP5	gastric_reflux	347166	9	34295	0	0.9	0.00	0.64	0.54	0.00	4.32	2126	0.95
17q11.2_del_NF1	gastric_reflux	347165	8	34296	1	0.8	1.24	0.64	1.54	0.16	6.88	2131	0.95
TAR_dup	gastric_reflux	347526	369	34334	39	36.5	1.06	0.66	1.08	0.76	1.49	2172	0.95
16p11.2distal_dup	gastric_reflux	347271	114	34308	13	11.3	1.14	0.66	1.14	0.60	1.97	2181	0.95
15q13.3_del	gastric_reflux	347190	33	34299	4	3.3	1.20	0.67	1.24	0.40	3.03	2222	0.95
7q11.23_dup_distal	gastric_reflux	347179	22	34296	1	2.2	0.48	0.68	0.72	0.08	2.78	2236	0.95
13q12.12_dup	gastric_reflux	347349	192	34315	20	19.0	1.05	0.75	1.08	0.66	1.66	2439	0.97
2q21.1_dup	gastric_reflux	347207	50	34300	5	4.9	1.01	0.80	1.12	0.41	2.48	2601	0.97
TAR_del	gastric_reflux	347220	63	34301	6	6.2	0.97	0.84	1.09	0.44	2.27	2703	0.97
2q21.1_del	gastric_reflux	347194	37	34298	3	3.7	0.83	0.87	0.92	0.25	2.40	2797	0.98
NRXN1_del	gastric_reflux	347300	143	34309	14	14.1	0.99	0.88	1.04	0.58	1.73	2807	0.98
2q11.2_dup	gastric_reflux	347181	24	34297	2	2.4	0.86	0.89	1.10	0.22	3.39	2856	0.98
3q29_dup	gastric_reflux	347162	5	34295	0	0.49	0.00	0.91	1.19	0.01	10.80	2906	0.98
22q11.2distal_del	gastric_reflux	347162	5	34295	0	0.5	0.00	0.93	0.88	0.01	8.04	2958	0.99
Potocki_Lupski	gastric_reflux	347162	5	34295	0	0.5	0.00	0.95	0.92	0.01	8.13	3024	0.99
8p23.1_dup	gastric_reflux	347162	5	34295	0	0.5	0.00	0.96	0.93	0.01	8.20	3031	0.99
2q13_del	gastric_reflux	347204	47	34299	4	4.6	0.87	0.99	0.99	0.32	2.35	3097	1.00
15q11.2_del	gastric_reflux	348586	1429	34436	141	141.2	1.00	0.99	1.00	0.84	1.18	3099	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q21.1_del	gastrointestinal_ulcer	371136	36	10356	4	1.0	3.68	0.015	4.35	1.39	10.59	157	0.30
10q23_dup	gastrointestinal_ulcer	371106	6	10353	1	0.2	5.26	0.053	9.61	0.96	49.66	343	0.49
17p12_CMT1A_dup	gastrointestinal_ulcer	371210	110	10358	6	3.1	1.91	0.090	2.16	0.87	4.43	490	0.58
22q11.2_del	gastrointestinal_ulcer	371109	9	10353	1	0.3	3.68	0.11	5.76	0.60	26.30	549	0.62
16p11.2distal_dup	gastrointestinal_ulcer	371221	121	10358	6	3.4	1.74	0.11	2.04	0.83	4.17	562	0.63
13q12.12_del	gastrointestinal_ulcer	371184	84	10352	0	2.3	0.00	0.11	0.19	0.00	1.33	570	0.63
15q11q13del_BP3-BP4	gastrointestinal_ulcer	371113	13	10353	1	0.4	2.63	0.13	4.98	0.54	20.77	637	0.64
16p12.1_del	gastrointestinal_ulcer	371325	225	10362	10	6.3	1.57	0.14	1.67	0.84	2.95	656	0.65
1q21.1_del	gastrointestinal_ulcer	371201	101	10357	5	2.8	1.74	0.16	1.99	0.74	4.32	712	0.69
WBS_dup	gastrointestinal_ulcer	371112	12	10353	1	0.3	2.83	0.16	4.26	0.46	17.97	732	0.70
2q21.1_dup	gastrointestinal_ulcer	371152	52	10355	3	1.5	2.01	0.18	2.33	0.64	6.03	765	0.72
TAR_dup	gastrointestinal_ulcer	371493	393	10367	15	11.0	1.35	0.20	1.42	0.82	2.29	844	0.74
2q13_dup	gastrointestinal_ulcer	371168	68	10355	3	1.9	1.56	0.30	1.86	0.51	4.77	1170	0.81
16p11.2distal_del	gastrointestinal_ulcer	371154	54	10352	0	1.5	0.00	0.32	0.31	0.00	2.19	1223	0.81
16p11.2_del	gastrointestinal_ulcer	371198	98	10357	5	2.7	1.79	0.34	1.57	0.58	3.44	1289	0.83
17p12_HNPP_del	gastrointestinal_ulcer	371311	211	10360	8	5.9	1.35	0.36	1.40	0.65	2.62	1354	0.84
2q11.2_dup	gastrointestinal_ulcer	371125	25	10353	1	0.7	1.42	0.39	2.26	0.25	8.82	1446	0.85
15q13.3_del	gastrointestinal_ulcer	371135	35	10354	2	1.0	1.99	0.39	1.86	0.37	5.73	1455	0.85
15q11q13dup_BP3_BP4	gastrointestinal_ulcer	371148	48	10354	2	1.3	1.47	0.40	1.83	0.38	5.40	1465	0.85
3q29_dup	gastrointestinal_ulcer	371105	5	10352	0	0.14	0.00	0.41	4.42	0.03	40.57	1492	0.86
13q12del_CRYL1	gastrointestinal_ulcer	371457	357	10358	6	10.0	0.61	0.44	0.75	0.31	1.50	1593	0.87
16p12.1_dup	gastrointestinal_ulcer	371289	189	10355	3	5.3	0.58	0.46	0.68	0.19	1.70	1626	0.88
22q11.2_dup	gastrointestinal_ulcer	371357	257	10361	9	7.2	1.25	0.48	1.28	0.62	2.32	1696	0.88
8p23.1_dup	gastrointestinal_ulcer	371105	5	10352	0	0.1	0.00	0.49	3.30	0.03	29.57	1734	0.89
22q11.2distal_del	gastrointestinal_ulcer	371105	5	10352	0	0.1	0.00	0.50	3.19	0.02	29.69	1771	0.89
Potocki_Lupski	gastrointestinal_ulcer	371105	5	10352	0	0.1	0.00	0.52	3.05	0.02	28.22	1809	0.90
15q24_dup	gastrointestinal_ulcer	371108	8	10352	0	0.2	0.00	0.52	2.98	0.02	24.42	1821	0.90
10q11.2q11.23_del	gastrointestinal_ulcer	371154	54	10354	2	1.5	1.32	0.53	1.55	0.32	4.58	1839	0.90
2q11.2_del	gastrointestinal_ulcer	371128	28	10353	1	0.8	1.27	0.60	1.62	0.18	6.29	2001	0.93
10q11.2q11.23_dup	gastrointestinal_ulcer	371139	39	10353	1	1.1	0.92	0.64	1.52	0.17	5.75	2121	0.94
15q11q13dup_BP3_BP5	gastrointestinal_ulcer	371109	9	10352	0	0.3	0.00	0.68	1.93	0.01	15.89	2245	0.95
17q12_del	gastrointestinal_ulcer	371107	7	10352	0	0.2	0.00	0.72	1.79	0.01	15.28	2324	0.96
3q29_del	gastrointestinal_ulcer	371109	9	10352	0	0.3	0.00	0.72	1.75	0.01	14.08	2360	0.96
13q12dup_CRYL1	gastrointestinal_ulcer	371110	10	10352	0	0.3	0.00	0.75	1.65	0.01	12.97	2424	0.97
1q21.1_dup	gastrointestinal_ulcer	371263	163	10357	5	4.5	1.10	0.76	1.14	0.43	2.44	2476	0.96
PWS_dup	gastrointestinal_ulcer	371116	16	10352	0	0.4	0.00	0.78	1.53	0.01	11.62	2531	0.97
17q12_dup	gastrointestinal_ulcer	371196	96	10355	3	2.7	1.12	0.78	1.17	0.32	2.94	2542	0.97
2q13del(NPHP1)	gastrointestinal_ulcer	373370	2270	10412	60	63.3	0.95	0.79	0.97	0.74	1.24	2553	0.97
17q11.2_del_NF1	gastrointestinal_ulcer	371109	9	10352	0	0.3	0.00	0.79	1.50	0.01	11.99	2568	0.97
16p11.2_dup	gastrointestinal_ulcer	371227	127	10356	4	3.5	1.13	0.80	1.14	0.37	2.61	2576	0.97
15q13.3del(CHRNA7)	gastrointestinal_ulcer	371110	10	10352	0	0.3	0.00	0.81	1.43	0.01	11.54	2625	0.97
15q13.3dup(CHRNA7)	gastrointestinal_ulcer	373866	2766	10429	77	77.2	1.00	0.82	1.03	0.81	1.28	2631	0.97
TAR_del	gastrointestinal_ulcer	371168	68	10353	1	1.9	0.53	0.82	0.83	0.09	3.07	2640	0.97
16p13.11_dup	gastrointestinal_ulcer	371861	761	10374	22	21.2	1.04	0.82	1.05	0.67	1.56	2644	0.97
NRXN1_del	gastrointestinal_ulcer	371253	153	10356	4	4.3	0.94	0.87	1.08	0.36	2.47	2785	0.98
15q13.3_dup	gastrointestinal_ulcer	371318	218	10358	6	6.1	0.99	0.89	0.95	0.39	1.91	2839	0.98
2q13dup(NPHP1)	gastrointestinal_ulcer	372917	1817	10403	51	50.7	1.01	0.89	1.02	0.76	1.33	2840	0.98
16p13.11_del	gastrointestinal_ulcer	371221	121	10355	3	3.4	0.89	0.90	1.07	0.30	2.69	2878	0.98
7q11.23_dup_distal	gastrointestinal_ulcer	371123	23	10352	0	0.6	0.00	0.93	0.88	0.01	6.40	2943	0.98
22q11.2distal_dup	gastrointestinal_ulcer	371112	12	10352	0	0.3	0.00	0.93	1.14	0.01	8.93	2954	0.98
15q11.2_del	gastrointestinal_ulcer	372627	1527	10395	43	42.6	1.01	0.93	1.01	0.74	1.35	2962	0.99
13q12.12_dup	gastrointestinal_ulcer	371307	207	10357	5	5.8	0.87	0.94	0.97	0.36	2.05	2971	0.99
2q13_del	gastrointestinal_ulcer	371150	50	10353	1	1.4	0.72	0.97	1.03	0.12	3.85	3063	0.99
15q11.2_dup	gastrointestinal_ulcer	372965	1865	10404	52	52.0	1.00	0.99	1.00	0.75	1.30	3115	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
3q29_del	glaucoma	374931	7	6530	2	0.1	12.99	0.0016	23.02	4.14	90.99	47	0.10
PWS_dup	glaucoma	374938	14	6530	2	0.2	7.30	0.0033	16.99	3.09	62.56	67	0.15
22q11.2_dup	glaucoma	375181	257	6537	9	4.5	1.98	0.020	2.40	1.16	4.36	186	0.34
7q11.23_dup_distal	glaucoma	374945	21	6530	2	0.4	5.08	0.021	7.24	1.43	23.26	192	0.35
17q12_del	glaucoma	374930	6	6529	1	0.1	8.35	0.027	14.70	1.48	75.86	230	0.37
WBS_dup	glaucoma	374936	12	6529	1	0.2	4.49	0.069	7.68	0.81	34.20	399	0.54
17q12_dup	glaucoma	375019	95	6532	4	1.7	2.36	0.076	2.70	0.89	6.29	431	0.55
TAR_del	glaucoma	374990	66	6531	3	1.1	2.54	0.076	3.17	0.87	8.19	432	0.55
22q11.2distal_dup	glaucoma	374935	11	6529	1	0.2	4.87	0.087	6.54	0.70	28.03	477	0.57
16p13.11_del	glaucoma	375044	120	6532	4	2.1	1.88	0.13	2.27	0.75	5.23	648	0.64
13q12.12_del	glaucoma	375005	81	6531	3	1.4	2.09	0.17	2.34	0.64	5.96	751	0.72
16p13.11_dup	glaucoma	375699	775	6536	8	13.5	0.60	0.19	0.66	0.31	1.21	809	0.74
2q11.2_del	glaucoma	374952	28	6529	1	0.5	2.01	0.20	3.69	0.41	14.46	829	0.74
10q11.21q11.23_del	glaucoma	374978	54	6530	2	0.9	2.09	0.20	2.58	0.53	7.67	856	0.74
16p12.1_dup	glaucoma	375111	187	6533	5	3.3	1.52	0.25	1.72	0.64	3.66	1009	0.78
3q29_dup	glaucoma	374929	5	6528	0	0.09	0.00	0.26	8.95	0.07	86.01	1034	0.79
NRXN1_del	glaucoma	375077	153	6532	4	2.7	1.49	0.27	1.79	0.59	4.09	1047	0.80
10q11.21q11.23_dup	glaucoma	374963	39	6529	1	0.7	1.46	0.27	2.94	0.33	11.24	1057	0.80
22q11.2distal_del	glaucoma	374929	5	6528	0	0.1	0.00	0.27	8.38	0.06	81.24	1073	0.80
13q12.12_dup	glaucoma	375131	207	6533	5	3.6	1.38	0.30	1.62	0.61	3.44	1174	0.81
15q24_dup	glaucoma	374932	8	6528	0	0.1	0.00	0.34	5.97	0.05	51.83	1278	0.83
Potocki_Lupski	glaucoma	374929	5	6528	0	0.1	0.00	0.37	5.25	0.04	49.49	1363	0.85
16p11.2_del	glaucoma	375027	103	6528	0	1.8	0.00	0.41	0.37	0.00	2.59	1496	0.86
1q21.1_dup	glaucoma	375091	167	6529	1	2.9	0.35	0.42	0.55	0.06	1.98	1508	0.87
22q11.2_del	glaucoma	374934	10	6528	0	0.2	0.00	0.42	4.14	0.03	35.22	1536	0.87
15q11q13dup_BP3_BP5	glaucoma	374933	9	6528	0	0.2	0.00	0.43	4.00	0.03	33.64	1557	0.87
8p23.1_dup	glaucoma	374929	5	6528	0	0.1	0.00	0.44	4.00	0.03	37.13	1568	0.87
10q23_dup	glaucoma	374931	7	6528	0	0.1	0.00	0.44	3.95	0.03	37.08	1583	0.87
15q11.2_del	glaucoma	376463	1539	6559	31	26.8	1.15	0.44	1.15	0.79	1.61	1594	0.87
17p12_HNPP_del	glaucoma	375138	214	6533	5	3.7	1.33	0.46	1.40	0.53	2.98	1636	0.88
2q13_dup	glaucoma	374995	71	6528	0	1.2	0.00	0.47	0.42	0.00	2.89	1674	0.88
15q13.3del(CHRNA7)	glaucoma	374934	10	6528	0	0.2	0.00	0.48	3.40	0.03	27.85	1701	0.88
15q11q13dup_BP3_BP4	glaucoma	374973	49	6529	1	0.9	1.17	0.49	1.89	0.21	7.14	1726	0.89
13q12del_CRYL1	glaucoma	375280	356	6535	7	6.2	1.13	0.49	1.31	0.57	2.51	1729	0.89
15q13.3_dup	glaucoma	375146	222	6530	2	3.9	0.52	0.50	0.67	0.14	1.90	1761	0.89
13q12dup_CRYL1	glaucoma	374934	10	6528	0	0.2	0.00	0.52	3.03	0.02	24.90	1803	0.90
15q13.3dup(CHRNA7)	glaucoma	377724	2800	6571	43	48.7	0.88	0.54	0.91	0.66	1.22	1856	0.91
15q11q13del_BP3-BP4	glaucoma	374938	14	6528	0	0.2	0.00	0.58	2.49	0.02	19.18	1957	0.93
2q21.1_dup	glaucoma	374978	54	6529	1	0.9	1.06	0.59	1.62	0.18	6.06	1980	0.93
TAR_dup	glaucoma	375327	403	6533	5	7.0	0.72	0.61	0.81	0.30	1.70	2037	0.94
17q11.2_del_NF1	glaucoma	374933	9	6528	0	0.2	0.00	0.62	2.27	0.02	18.77	2057	0.94
16p11.2distal_dup	glaucoma	375049	125	6530	2	2.2	0.92	0.68	1.31	0.27	3.78	2248	0.95
16p11.2distal_del	glaucoma	374978	54	6528	0	0.9	0.00	0.70	0.60	0.00	4.24	2293	0.96
17p12_CMT1A_dup	glaucoma	375039	115	6529	1	2.0	0.50	0.72	0.75	0.09	2.73	2326	0.96
2q21.1_del	glaucoma	374964	40	6528	0	0.7	0.00	0.75	0.65	0.01	4.65	2431	0.97
2q13_del	glaucoma	374975	51	6528	0	0.9	0.00	0.76	0.66	0.01	4.68	2457	0.97
2q13del(NPHP1)	glaucoma	377216	2292	6566	38	39.9	0.95	0.80	0.96	0.68	1.30	2570	0.97
16p12.1_del	glaucoma	375156	232	6531	3	4.0	0.75	0.81	0.88	0.25	2.19	2620	0.97
16p11.2_dup	glaucoma	375053	129	6530	2	2.2	0.89	0.84	1.14	0.24	3.28	2694	0.97
2q13dup(NPHP1)	glaucoma	376761	1837	6559	31	32.0	0.97	0.85	0.97	0.66	1.35	2725	0.97
15q13.3_del	glaucoma	374961	37	6528	0	0.6	0.00	0.86	0.79	0.01	5.60	2766	0.97
2q11.2_dup	glaucoma	374950	26	6528	0	0.5	0.00	0.91	1.19	0.01	8.72	2886	0.98
1q21.1_del	glaucoma	375029	105	6529	1	1.8	0.55	0.91	0.91	0.10	3.31	2894	0.98
15q11.2_dup	glaucoma	376810	1886	6559	31	32.8	0.94	0.94	0.99	0.68	1.38	2998	0.99



CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2distal_del	gout	374410	48	7096	6	0.9	5.98	0.00046	6.50	2.52	14.40	34	0.042*
22q11.2_dup	gout	374628	266	7090	0	5.0	0.00	0.0071	0.09	0.00	0.63	97	0.23
16p11.2_del	gout	374459	97	7096	6	1.8	3.13	0.017	3.15	1.26	6.64	170	0.32
15q11.2_dup	gout	376231	1869	7138	48	35.5	1.35	0.032	1.40	1.03	1.84	253	0.39
16p11.2distal_dup	gout	374484	122	7095	5	2.3	2.12	0.057	2.63	0.97	5.76	357	0.50
17q12_del	gout	374368	6	7091	1	0.1	7.69	0.079	7.42	0.74	39.84	449	0.55
16p13.11_dup	gout	375124	762	7111	21	14.4	1.44	0.082	1.51	0.95	2.27	461	0.56
22q11.2distal_dup	gout	374373	11	7091	1	0.2	4.48	0.097	6.28	0.65	29.93	519	0.59
15q13.3del(CHRNA7)	gout	374371	9	7091	1	0.2	5.38	0.099	6.18	0.64	29.22	525	0.59
WBS_dup	gout	374374	12	7091	1	0.2	4.14	0.10	5.99	0.62	28.14	537	0.61
16p12.1_dup	gout	374548	186	7096	6	3.5	1.68	0.12	2.00	0.81	4.11	611	0.63
16p12.1_del	gout	374590	228	7097	7	4.3	1.60	0.15	1.82	0.79	3.56	680	0.67
15q13.3dup(CHRNA7)	gout	377143	2781	7152	62	52.7	1.17	0.16	1.21	0.93	1.55	709	0.69
17p12_CMT1A_dup	gout	374474	112	7094	4	2.1	1.86	0.17	2.11	0.69	4.96	745	0.71
15q13.3_del	gout	374397	35	7092	2	0.7	2.91	0.18	2.82	0.56	8.91	769	0.72
16p13.11_del	gout	374486	124	7090	0	2.3	0.00	0.19	0.24	0.00	1.67	820	0.74
2q11.2_dup	gout	374387	25	7091	1	0.5	2.07	0.20	3.71	0.40	15.18	835	0.75
10q11.21q11.23_del	gout	374416	54	7092	2	1.0	1.92	0.20	2.62	0.53	8.03	850	0.74
TAR_dup	gout	374759	397	7101	11	7.5	1.45	0.25	1.45	0.75	2.50	995	0.78
15q24_dup	gout	374370	8	7090	0	0.2	0.00	0.26	9.34	0.07	86.48	1017	0.79
2q13del(NPHP1)	gout	376657	2295	7125	35	43.4	0.81	0.26	0.83	0.58	1.14	1031	0.79
2q21.1_dup	gout	374415	53	7092	2	1.0	1.96	0.31	2.08	0.42	6.26	1212	0.81
22q11.2distal_del	gout	374367	5	7090	0	0.1	0.00	0.33	6.39	0.05	72.35	1253	0.82
3q29_dup	gout	374367	5	7090	0	0.09	0.00	0.38	5.04	0.04	47.25	1395	0.85
TAR_del	gout	374431	69	7090	0	1.3	0.00	0.41	0.37	0.00	2.62	1493	0.86
Potocki_Lupski	gout	374367	5	7090	0	0.1	0.00	0.42	4.39	0.03	52.25	1515	0.87
2q13dup(NPHP1)	gout	376190	1828	7130	40	34.6	1.15	0.45	1.13	0.81	1.53	1614	0.87
10q11.21q11.23_dup	gout	374401	39	7091	1	0.7	1.35	0.45	2.02	0.22	7.78	1618	0.87
15q11q13dup_BP3_BP5	gout	374371	9	7090	0	0.2	0.00	0.48	3.50	0.03	32.38	1685	0.88
13q12dup_CRYL1	gout	374372	10	7090	0	0.2	0.00	0.48	3.42	0.03	32.83	1717	0.88
8p23.1_dup	gout	374367	5	7090	0	0.1	0.00	0.49	3.41	0.03	34.87	1721	0.88
17q12_dup	gout	374458	96	7093	3	1.8	1.63	0.52	1.45	0.40	3.70	1826	0.90
2q13_del	gout	374413	51	7090	0	1.0	0.00	0.53	0.46	0.00	3.25	1841	0.90
17p12_HNPP_del	gout	374576	214	7095	5	4.1	1.23	0.55	1.31	0.49	2.82	1883	0.91
10q23_dup	gout	374369	7	7090	0	0.1	0.00	0.56	2.66	0.02	24.70	1929	0.91
3q29_del	gout	374371	9	7090	0	0.2	0.00	0.61	2.35	0.02	20.56	2027	0.94
15q11q13del_BP3-BP4	gout	374376	14	7090	0	0.3	0.00	0.62	2.24	0.02	17.91	2067	0.94
1q21.1_del	gout	374466	104	7092	2	2.0	1.02	0.62	1.40	0.29	4.10	2068	0.94
PWS_dup	gout	374378	16	7090	0	0.3	0.00	0.63	2.23	0.02	18.68	2080	0.94
22q11.2_del	gout	374372	10	7090	0	0.2	0.00	0.64	2.16	0.02	18.41	2118	0.94
16p11.2_dup	gout	374490	128	7093	3	2.4	1.23	0.65	1.30	0.36	3.29	2148	0.95
2q21.1_del	gout	374402	40	7090	0	0.8	0.00	0.66	0.57	0.00	4.09	2184	0.95
15q11q13dup_BP3_BP4	gout	374411	49	7091	1	0.9	1.08	0.70	1.41	0.16	5.36	2283	0.96
17q11.2_del_NF1	gout	374371	9	7090	0	0.2	0.00	0.71	1.83	0.01	15.99	2306	0.96
15q13.3_dup	gout	374582	220	7094	4	4.2	0.96	0.72	0.85	0.28	1.93	2365	0.96
13q12del_CRYL1	gout	374720	358	7095	5	6.8	0.74	0.74	0.87	0.33	1.84	2410	0.96
NRXN1_del	gout	374517	155	7092	2	2.9	0.69	0.78	0.84	0.17	2.41	2512	0.97
2q11.2_del	gout	374391	29	7090	0	0.5	0.00	0.81	0.72	0.01	5.31	2608	0.97
1q21.1_dup	gout	374527	165	7093	3	3.1	0.96	0.83	1.12	0.31	2.83	2682	0.97
13q12.12_del	gout	374445	83	7091	1	1.6	0.64	0.84	0.85	0.10	3.17	2717	0.97
7q11.23_dup_distal	gout	374385	23	7090	0	0.4	0.00	0.90	1.21	0.01	9.35	2870	0.98
13q12.12_dup	gout	374571	209	7093	3	4.0	0.76	0.90	0.93	0.26	2.34	2873	0.98
15q11.2_del	gout	375902	1540	7120	30	29.2	1.03	0.92	0.98	0.67	1.38	2915	0.98
2q13_dup	gout	374432	70	7091	1	1.3	0.76	0.99	1.01	0.11	3.78	3106	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p12.1_del	heart_other	369848	217	11839	18	6.9	2.47	0.00029	2.77	1.65	4.36	27	0.034*
1q21.1_dup	heart_other	369787	156	11833	12	5.0	2.30	0.0056	2.55	1.35	4.39	82	0.21
13q12dup_CRYL1	heart_other	369639	8	11823	2	0.3	6.45	0.0091	11.24	2.05	42.70	114	0.25
16p11.2distal_dup	heart_other	369749	118	11830	9	3.8	2.29	0.0091	2.77	1.32	5.15	115	0.25
15q24_dup	heart_other	369638	7	11822	1	0.2	4.03	0.034	12.25	1.28	57.33	258	0.41
16p11.2_dup	heart_other	369754	123	11829	8	3.9	1.97	0.067	2.07	0.95	3.94	383	0.55
16p11.2distal_del	heart_other	369681	50	11825	4	1.6	2.39	0.070	2.81	0.91	6.72	403	0.54
16p12.1_dup	heart_other	369813	182	11831	10	5.8	1.68	0.073	1.87	0.94	3.35	422	0.54
1q21.1_del	heart_other	369731	100	11827	6	3.2	1.83	0.084	2.20	0.89	4.55	467	0.56
10q11.21q11.23_del	heart_other	369683	52	11825	4	1.7	2.30	0.091	2.60	0.84	6.25	493	0.58
NRXN1_del	heart_other	369780	149	11829	8	4.8	1.64	0.11	1.86	0.85	3.51	561	0.63
3q29_del	heart_other	369639	8	11822	1	0.3	3.59	0.12	5.41	0.56	25.38	602	0.63
15q13.3del(CHRNA7)	heart_other	369640	9	11822	1	0.3	3.23	0.15	4.64	0.49	21.17	691	0.68
22q11.2_dup	heart_other	369885	254	11833	12	8.1	1.46	0.16	1.55	0.83	2.65	717	0.69
15q11q13dup_BP3_BP4	heart_other	369678	47	11824	3	1.5	1.94	0.16	2.41	0.66	6.32	728	0.70
16p13.11_del	heart_other	369749	118	11827	6	3.8	1.56	0.17	1.85	0.75	3.81	736	0.70
15q13.3dup(CHRNA7)	heart_other	372375	2744	11920	99	87.8	1.12	0.18	1.15	0.94	1.40	762	0.72
22q11.2distal_dup	heart_other	369642	11	11822	1	0.4	2.69	0.23	3.44	0.37	14.80	920	0.77
16p11.2_del	heart_other	369729	98	11826	5	3.1	1.57	0.26	1.72	0.63	3.75	1025	0.79
16p13.11_dup	heart_other	370385	754	11850	29	24.1	1.20	0.27	1.24	0.84	1.76	1074	0.80
2q21.1_dup	heart_other	369683	52	11824	3	1.7	1.76	0.27	1.95	0.53	5.09	1075	0.80
17q12_dup	heart_other	369725	94	11826	5	3.0	1.63	0.31	1.62	0.60	3.52	1191	0.81
13q12.12_del	heart_other	369714	83	11822	1	2.7	0.38	0.37	0.52	0.06	1.89	1382	0.85
13q12.12_dup	heart_other	369835	204	11829	8	6.5	1.22	0.39	1.37	0.63	2.57	1459	0.85
7q11.23_dup_distal	heart_other	369653	22	11822	1	0.7	1.40	0.40	2.25	0.25	8.99	1462	0.85
3q29_dup	heart_other	369636	5	11821	0	0.16	0.00	0.42	4.26	0.03	38.56	1511	0.87
TAR_dup	heart_other	370024	393	11836	15	12.6	1.19	0.44	1.23	0.71	1.98	1582	0.87
2q13_del	heart_other	369680	49	11823	2	1.6	1.27	0.47	1.66	0.34	4.96	1669	0.88
22q11.2distal_del	heart_other	369636	5	11821	0	0.2	0.00	0.48	3.48	0.03	33.21	1695	0.88
10q11.21q11.23_dup	heart_other	369671	40	11821	0	1.3	0.00	0.53	0.46	0.00	3.25	1843	0.90
Potocki_Lupski	heart_other	369636	5	11821	0	0.2	0.00	0.56	2.72	0.02	25.87	1911	0.91
2q13dup(NPHP1)	heart_other	371445	1814	11875	54	58.0	0.93	0.56	0.92	0.70	1.20	1926	0.92
2q11.2_del	heart_other	369660	29	11821	0	0.9	0.00	0.60	0.51	0.00	3.72	2013	0.94
8p23.1_dup	heart_other	369636	5	11821	0	0.2	0.00	0.62	2.25	0.02	20.81	2072	0.94
15q11.2_del	heart_other	371148	1517	11874	53	48.5	1.09	0.65	1.07	0.80	1.39	2150	0.94
15q11q13dup_BP3_BP5	heart_other	369640	9	11821	0	0.3	0.00	0.67	2.01	0.02	16.65	2196	0.95
17p12_CMT1A_dup	heart_other	369743	112	11825	4	3.6	1.11	0.67	1.24	0.41	2.86	2201	0.95
10q23_dup	heart_other	369638	7	11821	0	0.2	0.00	0.69	1.89	0.01	17.33	2280	0.95
13q12del_CRYL1	heart_other	369983	352	11832	11	11.3	0.98	0.72	1.12	0.58	1.92	2349	0.96
2q11.2_dup	heart_other	369657	26	11821	0	0.8	0.00	0.76	0.66	0.01	4.78	2451	0.97
17q12_del	heart_other	369638	7	11821	0	0.2	0.00	0.76	1.61	0.01	14.05	2472	0.97
15q13.3_dup	heart_other	369847	216	11829	8	6.9	1.15	0.78	1.11	0.51	2.07	2523	0.97
22q11.2_del	heart_other	369641	10	11821	0	0.3	0.00	0.79	1.53	0.01	12.75	2546	0.97
TAR_del	heart_other	369698	67	11823	2	2.1	0.94	0.79	1.19	0.25	3.51	2558	0.97
15q11q13del_BP3-BP4	heart_other	369645	14	11821	0	0.4	0.00	0.84	1.36	0.01	10.34	2704	0.97
17p12_HNPP_del	heart_other	369844	213	11827	6	6.8	0.88	0.84	0.92	0.38	1.87	2718	0.97
PWS_dup	heart_other	369647	16	11821	0	0.5	0.00	0.85	1.33	0.01	10.49	2739	0.97
2q13del(NPHP1)	heart_other	371888	2257	11894	73	72.2	1.01	0.85	1.02	0.80	1.28	2740	0.97
2q13_dup	heart_other	369700	69	11823	2	2.2	0.91	0.89	1.10	0.23	3.21	2837	0.98
17q11.2_del_NF1	heart_other	369640	9	11821	0	0.3	0.00	0.90	1.22	0.01	9.99	2868	0.98
WBS_dup	heart_other	369644	13	11821	0	0.4	0.00	0.92	1.16	0.01	9.06	2931	0.98
15q13.3_del	heart_other	369667	36	11822	1	1.2	0.87	0.92	1.09	0.12	4.16	2932	0.98
2q21.1_del	heart_other	369670	39	11822	1	1.2	0.81	0.93	1.08	0.12	4.09	2952	0.98
15q11.2_dup	heart_other	371489	1858	11880	59	59.4	0.99	0.94	1.01	0.77	1.30	2977	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
15q13.3del(CHRNA7)	heart_valve	373084	8	8378	2	0.2	9.11	0.0046	14.45	2.69	53.98	77	0.19
16p11.2distal_dup	heart_valve	373196	120	8383	7	2.7	2.51	0.011	3.06	1.33	6.00	131	0.27
22q11.2distal_del	heart_valve	373080	4	8377	1	0.1	9.11	0.018	19.46	1.89	110.43	174	0.33
15q13.3dup(CHRNA7)	heart_valve	375839	2763	8456	80	62.2	1.28	0.020	1.32	1.05	1.64	177	0.35
Potocki_Lupski	heart_valve	373080	4	8377	1	0.1	9.11	0.027	14.81	1.46	82.01	232	0.37
10q23_dup	heart_valve	373082	6	8377	1	0.1	6.51	0.039	11.66	1.17	60.16	286	0.43
22q11.2_del	heart_valve	373085	9	8377	1	0.2	4.55	0.053	9.22	0.97	41.69	338	0.49
13q12dup_CRYL1	heart_valve	373085	9	8377	1	0.2	4.55	0.073	7.40	0.78	32.91	418	0.55
10q11.21q11.23_dup	heart_valve	373114	38	8378	2	0.9	2.28	0.10	3.57	0.73	10.75	534	0.60
13q12.12_del	heart_valve	373156	80	8380	4	1.8	2.17	0.11	2.38	0.78	5.53	572	0.63
22q11.2distal_dup	heart_valve	373087	11	8377	1	0.2	3.80	0.13	4.94	0.53	20.98	643	0.64
1q21.1_del	heart_valve	373178	102	8380	4	2.3	1.72	0.17	2.09	0.69	4.81	750	0.71
17q12_dup	heart_valve	373171	95	8380	4	2.1	1.84	0.18	2.05	0.67	4.73	783	0.73
13q12.12_dup	heart_valve	373281	205	8383	7	4.6	1.50	0.19	1.71	0.75	3.30	798	0.73
15q11q13dup_BP3_BP4	heart_valve	373124	48	8378	2	1.1	1.82	0.23	2.44	0.50	7.23	927	0.77
7q11.23_dup_distal	heart_valve	373098	22	8377	1	0.5	1.98	0.24	3.23	0.36	12.67	957	0.78
13q12del_CRYL1	heart_valve	373435	359	8380	4	8.1	0.50	0.24	0.60	0.20	1.35	979	0.78
16p11.2distal_del	heart_valve	373128	52	8378	2	1.2	1.69	0.26	2.26	0.47	6.66	1039	0.79
NRXN1_del	heart_valve	373228	152	8381	5	3.4	1.45	0.28	1.67	0.62	3.56	1078	0.80
16p13.11_dup	heart_valve	373838	762	8397	21	17.1	1.22	0.28	1.28	0.81	1.92	1092	0.80
2q11.2_dup	heart_valve	373101	25	8377	1	0.6	1.75	0.28	2.84	0.32	11.04	1115	0.80
16p13.11_del	heart_valve	373196	120	8380	4	2.7	1.47	0.30	1.72	0.57	3.94	1155	0.81
10q11.21q11.23_del	heart_valve	373130	54	8378	2	1.2	1.63	0.33	2.01	0.41	5.92	1257	0.82
3q29_dup	heart_valve	373081	5	8376	0	0.11	0.00	0.35	5.61	0.04	51.88	1320	0.84
16p12.1_dup	heart_valve	373266	190	8378	2	4.3	0.47	0.37	0.59	0.12	1.68	1373	0.85
TAR_del	heart_valve	373143	67	8378	2	1.5	1.32	0.43	1.74	0.36	5.09	1545	0.87
15q24_dup	heart_valve	373084	8	8376	0	0.2	0.00	0.45	3.82	0.03	31.69	1599	0.87
15q11.2_del	heart_valve	374616	1540	8406	30	34.6	0.87	0.45	0.87	0.60	1.23	1615	0.87
2q21.1_dup	heart_valve	373131	55	8376	0	1.2	0.00	0.46	0.41	0.00	2.82	1629	0.88
17p12_CMT1A_dup	heart_valve	373191	115	8377	1	2.6	0.39	0.48	0.59	0.07	2.13	1704	0.88
8p23.1_dup	heart_valve	373081	5	8376	0	0.1	0.00	0.48	3.40	0.03	30.60	1710	0.88
2q13dup(NPHP1)	heart_valve	374907	1831	8413	37	41.1	0.90	0.52	0.90	0.64	1.23	1828	0.90
15q13.3_del	heart_valve	373112	36	8377	1	0.8	1.23	0.54	1.73	0.19	6.56	1872	0.91
17q12_del	heart_valve	373083	7	8376	0	0.2	0.00	0.55	2.75	0.02	23.35	1885	0.91
15q11q13dup_BP3_BP5	heart_valve	373085	9	8376	0	0.2	0.00	0.56	2.65	0.02	21.69	1919	0.91
3q29_del	heart_valve	373085	9	8376	0	0.2	0.00	0.59	2.41	0.02	19.44	1995	0.93
2q21.1_del	heart_valve	373116	40	8376	0	0.9	0.00	0.61	0.52	0.00	3.66	2030	0.94
2q13del(NPHP1)	heart_valve	375351	2275	8431	55	51.1	1.08	0.63	1.07	0.81	1.38	2109	0.94
2q13_del	heart_valve	373126	50	8377	1	1.1	0.89	0.66	1.47	0.17	5.48	2178	0.95
PWS_dup	heart_valve	373092	16	8376	0	0.4	0.00	0.68	1.91	0.01	14.58	2251	0.95
17q11.2_del_NF1	heart_valve	373085	9	8376	0	0.2	0.00	0.68	1.91	0.01	15.34	2252	0.95
22q11.2_dup	heart_valve	373336	260	8382	6	5.8	1.03	0.69	1.18	0.48	2.37	2261	0.95
15q11q13del_BP3-BP4	heart_valve	373090	14	8376	0	0.3	0.00	0.70	1.84	0.01	13.90	2291	0.95
15q11.2_dup	heart_valve	374953	1877	8416	40	42.1	0.95	0.72	0.94	0.68	1.28	2350	0.96
16p11.2_dup	heart_valve	373204	128	8379	3	2.9	1.04	0.73	1.21	0.34	3.04	2375	0.96
WBS_dup	heart_valve	373089	13	8376	0	0.3	0.00	0.73	1.71	0.01	13.17	2384	0.96
16p11.2_del	heart_valve	373177	101	8378	2	2.3	0.88	0.76	1.23	0.25	3.53	2461	0.97
16p12.1_del	heart_valve	373307	231	8380	4	5.2	0.78	0.79	0.88	0.29	2.00	2565	0.97
17p12_HNPP_del	heart_valve	373290	214	8381	5	4.8	1.04	0.81	1.11	0.42	2.35	2618	0.97
15q13.3_dup	heart_valve	373295	219	8381	5	4.9	1.02	0.82	1.10	0.41	2.34	2648	0.97
2q11.2_del	heart_valve	373105	29	8376	0	0.7	0.00	0.88	0.82	0.01	5.86	2821	0.98
TAR_dup	heart_valve	373476	400	8384	8	9.0	0.89	0.91	0.96	0.45	1.78	2903	0.98
1q21.1_dup	heart_valve	373241	165	8379	3	3.7	0.81	0.95	0.97	0.27	2.41	3017	0.99
2q13_dup	heart_valve	373146	70	8377	1	1.6	0.64	0.97	0.97	0.11	3.56	3069	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
1q21.1_del	heartfailure	376477	100	5081	6	1.3	4.25	0.0011	5.34	2.14	11.15	40	0.083*
17p12_CMT1A_dup	heartfailure	376487	110	5081	6	1.5	3.89	0.0034	4.32	1.73	9.03	68	0.16
16p11.2_del	heartfailure	376474	97	5081	6	1.3	4.38	0.0045	4.16	1.64	8.84	76	0.19
22q11.2_dup	heartfailure	376634	257	5084	9	3.5	2.54	0.011	2.64	1.27	4.83	133	0.27
10q11.21q11.23_del	heartfailure	376430	53	5078	3	0.7	4.03	0.028	4.53	1.21	12.15	237	0.37
3q29_del	heartfailure	376385	8	5076	1	0.1	8.35	0.034	12.56	1.28	61.19	259	0.41
22q11.2_del	heartfailure	376386	9	5076	1	0.1	7.52	0.038	11.90	1.19	59.49	281	0.43
22q11.2distal_dup	heartfailure	376388	11	5076	1	0.1	6.26	0.068	7.81	0.82	34.90	390	0.54
16p12.1_del	heartfailure	376606	229	5081	6	3.1	1.92	0.092	2.13	0.87	4.33	498	0.58
3q29_dup	heartfailure	376382	5	5075	0	0.07	0.00	0.21	12.97	0.10	118.28	866	0.75
15q24_dup	heartfailure	376385	8	5075	0	0.1	0.00	0.23	10.43	0.08	86.73	946	0.78
22q11.2distal_del	heartfailure	376382	5	5075	0	0.1	0.00	0.30	7.43	0.06	75.31	1146	0.81
16p13.11_del	heartfailure	376501	124	5075	0	1.7	0.00	0.32	0.32	0.00	2.19	1224	0.81
15q13.3_dup	heartfailure	376596	219	5080	5	3.0	1.68	0.34	1.56	0.58	3.34	1285	0.82
Potocki_Lupski	heartfailure	376382	5	5075	0	0.1	0.00	0.34	6.10	0.04	62.82	1286	0.82
15q11.2_dup	heartfailure	378264	1887	5105	30	25.5	1.18	0.34	1.20	0.82	1.69	1291	0.83
8p23.1_dup	heartfailure	376382	5	5075	0	0.1	0.00	0.34	5.86	0.04	55.68	1302	0.83
13q12del_CRYL1	heartfailure	376738	361	5077	2	4.9	0.41	0.37	0.59	0.12	1.67	1359	0.85
16p13.11_dup	heartfailure	377147	770	5088	13	10.4	1.25	0.37	1.30	0.72	2.14	1362	0.85
15q11q13dup_BP3_BP5	heartfailure	376386	9	5075	0	0.1	0.00	0.39	4.73	0.04	40.87	1443	0.85
13q12dup_CRYL1	heartfailure	376387	10	5075	0	0.1	0.00	0.41	4.34	0.03	36.56	1498	0.86
10q23_dup	heartfailure	376384	7	5075	0	0.1	0.00	0.43	4.12	0.03	40.46	1551	0.87
15q11q13del_BP3-BP4	heartfailure	376391	14	5075	0	0.2	0.00	0.44	3.89	0.03	29.85	1570	0.87
PWS_dup	heartfailure	376393	16	5075	0	0.2	0.00	0.47	3.52	0.03	29.04	1665	0.88
1q21.1_dup	heartfailure	376542	165	5078	3	2.2	1.34	0.47	1.52	0.42	3.82	1677	0.88
17q12_del	heartfailure	376384	7	5075	0	0.1	0.00	0.48	3.44	0.03	31.02	1703	0.88
16p12.1_dup	heartfailure	376568	191	5076	1	2.6	0.39	0.49	0.59	0.07	2.14	1724	0.88
16p11.2distal_dup	heartfailure	376502	125	5077	2	1.7	1.18	0.50	1.60	0.33	4.62	1755	0.89
17q11.2_del_NF1	heartfailure	376386	9	5075	0	0.1	0.00	0.53	2.91	0.02	24.34	1838	0.90
15q13.3del(CHRNA7)	heartfailure	376387	10	5075	0	0.1	0.00	0.54	2.82	0.02	23.68	1859	0.91
WBS_dup	heartfailure	376390	13	5075	0	0.2	0.00	0.56	2.68	0.02	21.28	1903	0.92
TAR_del	heartfailure	376445	68	5076	1	0.9	1.09	0.58	1.64	0.18	6.16	1962	0.93
2q13dup(NPHP1)	heartfailure	378218	1841	5102	27	24.8	1.09	0.64	1.10	0.73	1.57	2122	0.94
2q13_dup	heartfailure	376447	70	5076	1	0.9	1.06	0.65	1.50	0.17	5.59	2145	0.95
NRXN1_del	heartfailure	376533	156	5076	1	2.1	0.48	0.68	0.72	0.08	2.62	2223	0.95
TAR_dup	heartfailure	376779	402	5081	6	5.4	1.11	0.68	1.18	0.49	2.38	2247	0.95
2q21.1_dup	heartfailure	376432	55	5075	0	0.7	0.00	0.70	0.60	0.00	4.25	2281	0.96
16p11.2_dup	heartfailure	376507	130	5076	1	1.8	0.57	0.70	0.74	0.08	2.71	2304	0.96
7q11.23_dup_distal	heartfailure	376400	23	5075	0	0.3	0.00	0.72	1.77	0.01	13.33	2332	0.96
2q11.2_dup	heartfailure	376403	26	5075	0	0.4	0.00	0.73	1.72	0.01	12.66	2371	0.96
16p11.2distal_del	heartfailure	376431	54	5075	0	0.7	0.00	0.76	0.66	0.01	4.67	2459	0.97
15q13.3_del	heartfailure	376414	37	5075	0	0.5	0.00	0.79	0.69	0.01	5.04	2551	0.97
2q13_del	heartfailure	376428	51	5075	0	0.7	0.00	0.79	0.70	0.01	5.01	2566	0.97
17p12_HNPP_del	heartfailure	376593	216	5078	3	2.9	1.03	0.80	1.15	0.32	2.86	2597	0.97
13q12.12_del	heartfailure	376460	83	5076	1	1.1	0.89	0.83	1.21	0.14	4.46	2659	0.97
15q11q13dup_BP3_BP4	heartfailure	376427	50	5075	0	0.7	0.00	0.84	0.76	0.01	5.38	2707	0.97
2q21.1_del	heartfailure	376417	40	5075	0	0.5	0.00	0.87	0.80	0.01	5.76	2796	0.98
15q11.2_del	heartfailure	377925	1548	5097	22	20.9	1.05	0.91	1.02	0.65	1.52	2898	0.98
13q12.12_dup	heartfailure	376587	210	5077	2	2.8	0.71	0.91	0.93	0.19	2.66	2907	0.98
15q13.3dup(CHRNA7)	heartfailure	379184	2807	5111	36	37.8	0.95	0.94	0.99	0.70	1.35	2963	0.99
10q11.21q11.23_dup	heartfailure	376417	40	5075	0	0.5	0.00	0.94	1.12	0.01	8.14	2973	0.99
2q11.2_del	heartfailure	376406	29	5075	0	0.4	0.00	0.94	1.12	0.01	8.26	2980	0.99
2q13del(NPHP1)	heartfailure	378677	2300	5105	30	31.0	0.97	0.94	0.99	0.67	1.39	2986	0.99
17q12_dup	heartfailure	376475	98	5076	1	1.3	0.76	0.95	0.95	0.11	3.47	3001	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
PWS_dup	hepatic	372713	14	8755	2	0.3	5.45	0.018	7.79	1.53	25.71	173	0.33
1q21.1_dup	hepatic	372858	159	8762	9	3.7	2.33	0.023	2.37	1.14	4.32	206	0.35
3q29_dup	hepatic	372703	4	8754	1	0.09	8.72	0.025	15.80	1.56	88.19	215	0.36
Potocki_Lupski	hepatic	372703	4	8754	1	0.1	8.72	0.034	12.84	1.28	70.55	261	0.41
10q23_dup	hepatic	372705	6	8754	1	0.1	6.23	0.051	9.60	0.98	47.19	333	0.48
15q24_dup	hepatic	372706	7	8754	1	0.2	5.45	0.052	9.32	0.98	43.25	336	0.48
TAR_del	hepatic	372764	65	8757	4	1.5	2.53	0.053	3.00	0.98	7.01	342	0.49
2q11.2_dup	hepatic	372723	24	8755	2	0.6	3.35	0.061	4.57	0.92	14.12	365	0.52
16p13.11_del	hepatic	372817	118	8759	6	2.8	2.11	0.061	2.35	0.95	4.78	369	0.52
17q12_del	hepatic	372705	6	8754	1	0.1	6.23	0.075	7.45	0.77	36.18	429	0.55
16p11.2_del	hepatic	372796	97	8759	6	2.3	2.54	0.077	2.24	0.91	4.61	435	0.55
17q12_dup	hepatic	372798	99	8753	0	2.3	0.00	0.12	0.19	0.00	1.34	584	0.63
2q13dup(NPHP1)	hepatic	374513	1814	8807	54	42.7	1.26	0.12	1.25	0.94	1.63	601	0.63
16p11.2distal_del	hepatic	372750	51	8756	3	1.2	2.42	0.13	2.63	0.72	6.77	623	0.64
TAR_dup	hepatic	373093	394	8767	14	9.3	1.50	0.13	1.54	0.87	2.50	650	0.64
NRXN1_del	hepatic	372855	156	8754	1	3.7	0.28	0.20	0.41	0.05	1.46	831	0.75
2q21.1_del	hepatic	372737	38	8755	2	0.9	2.18	0.20	2.60	0.53	7.75	848	0.74
7q11.23_dup_distal	hepatic	372721	22	8754	1	0.5	1.89	0.25	3.07	0.34	11.94	1013	0.79
15q11.2_dup	hepatic	374564	1865	8805	52	43.8	1.18	0.26	1.18	0.88	1.53	1028	0.79
16p11.2_dup	hepatic	372825	126	8758	5	3.0	1.66	0.27	1.69	0.63	3.62	1046	0.80
15q13.3_dup	hepatic	372915	216	8761	8	5.1	1.56	0.29	1.48	0.69	2.76	1132	0.81
16p12.1_dup	hepatic	372889	190	8755	2	4.5	0.45	0.31	0.56	0.12	1.58	1189	0.82
13q12.12_del	hepatic	372780	81	8756	3	1.9	1.56	0.33	1.79	0.49	4.51	1256	0.82
2q13_del	hepatic	372748	49	8755	2	1.2	1.71	0.34	1.99	0.41	5.85	1287	0.82
15q11.2_del	hepatic	374238	1539	8784	31	36.1	0.86	0.37	0.86	0.59	1.20	1386	0.85
8p23.1_dup	hepatic	372704	5	8753	0	0.1	0.00	0.44	3.94	0.03	34.86	1573	0.87
16p12.1_del	hepatic	372927	228	8760	7	5.4	1.30	0.44	1.35	0.59	2.60	1587	0.87
17p12_HNPP_del	hepatic	372915	216	8756	3	5.1	0.60	0.45	0.68	0.19	1.69	1621	0.88
16p13.11_dup	hepatic	373467	768	8768	15	18.0	0.83	0.47	0.83	0.48	1.33	1659	0.88
2q11.2_del	hepatic	372727	28	8754	1	0.7	1.50	0.48	1.93	0.22	7.40	1692	0.88
13q12.12_dup	hepatic	372905	206	8759	6	4.8	1.23	0.49	1.33	0.55	2.68	1742	0.89
22q11.2distal_del	hepatic	372704	5	8753	0	0.1	0.00	0.50	3.17	0.02	28.68	1773	0.89
2q13_dup	hepatic	372768	69	8755	2	1.6	1.23	0.54	1.52	0.32	4.41	1860	0.91
1q21.1_del	hepatic	372802	103	8756	3	2.4	1.23	0.56	1.40	0.39	3.52	1905	0.91
10q11.21q11.23_dup	hepatic	372738	39	8754	1	0.9	1.09	0.58	1.64	0.18	6.15	1953	0.93
17q11.2_del_NF1	hepatic	372708	9	8753	0	0.2	0.00	0.65	2.06	0.02	16.34	2160	0.95
15q11q13dup_BP3_BP5	hepatic	372708	9	8753	0	0.2	0.00	0.66	2.05	0.02	16.37	2169	0.95
13q12dup_CRYL1	hepatic	372709	10	8753	0	0.2	0.00	0.67	2.00	0.02	15.55	2194	0.95
3q29_del	hepatic	372708	9	8753	0	0.2	0.00	0.67	1.97	0.02	15.61	2215	0.95
17p12_CMT1A_dup	hepatic	372812	113	8756	3	2.7	1.13	0.69	1.25	0.35	3.14	2265	0.95
15q13.3_del	hepatic	372735	36	8754	1	0.8	1.18	0.73	1.35	0.15	5.08	2391	0.96
22q11.2_del	hepatic	372709	10	8753	0	0.2	0.00	0.75	1.64	0.01	12.96	2440	0.97
15q13.3del(CHRNA7)	hepatic	372709	10	8753	0	0.2	0.00	0.75	1.63	0.01	12.89	2446	0.96
16p11.2distal_dup	hepatic	372824	125	8755	2	2.9	0.69	0.76	0.82	0.17	2.35	2450	0.97
22q11.2_dup	hepatic	372958	259	8760	7	6.1	1.15	0.76	1.12	0.49	2.16	2465	0.97
15q11q13del_BP3-BP4	hepatic	372713	14	8753	0	0.3	0.00	0.78	1.54	0.01	11.62	2524	0.97
15q11q13dup_BP3_BP4	hepatic	372748	49	8754	1	1.2	0.87	0.80	1.24	0.14	4.57	2598	0.97
WBS_dup	hepatic	372712	13	8753	0	0.3	0.00	0.83	1.39	0.01	10.59	2663	0.97
22q11.2distal_dup	hepatic	372711	12	8753	0	0.3	0.00	0.83	1.39	0.01	10.74	2666	0.97
2q21.1_dup	hepatic	372753	54	8754	1	1.3	0.79	0.90	1.12	0.13	4.11	2867	0.98
13q12del_CRYL1	hepatic	373055	356	8760	7	8.4	0.84	0.91	0.96	0.42	1.84	2895	0.98
2q13del(NPHP1)	hepatic	374976	2277	8806	53	53.5	0.99	0.96	1.01	0.76	1.31	3027	0.99
10q11.21q11.23_del	hepatic	372754	55	8754	1	1.3	0.78	0.97	1.03	0.12	3.79	3074	0.99
15q13.3dup(CHRNA7)	hepatic	375478	2779	8817	64	65.3	0.98	0.98	1.00	0.77	1.27	3081	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
22q11.2_dup	hernia	333779	215	47939	51	30.9	1.53	0.0013	1.71	1.24	2.32	45	0.094*
16p11.2_del	hernia	333645	81	47910	22	11.6	1.70	0.0084	1.99	1.20	3.16	105	0.25
17p12_HNPP_del	hernia	333767	203	47904	16	29.1	0.58	0.0089	0.54	0.31	0.86	111	0.25
16p12.1_del	hernia	333758	194	47929	41	27.9	1.39	0.020	1.53	1.07	2.13	181	0.35
TAR_dup	hernia	333904	340	47956	68	48.8	1.33	0.020	1.39	1.05	1.80	185	0.34
15q11q13dup_BP3_BP4	hernia	333603	39	47899	11	5.6	1.75	0.045	2.08	1.02	3.94	312	0.46
17q12_del	hernia	333568	4	47891	3	0.6	3.41	0.054	4.62	0.97	20.71	347	0.48
16p11.2_dup	hernia	333671	107	47912	24	15.4	1.46	0.068	1.55	0.97	2.38	393	0.54
2q13_dup	hernia	333631	67	47892	4	9.6	0.45	0.070	0.45	0.15	1.06	404	0.54
7q11.23_dup_distal	hernia	333587	23	47888	0	3.3	0.00	0.071	0.15	0.00	1.12	410	0.54
2q21.1_del	hernia	333595	31	47897	9	4.5	1.79	0.087	1.99	0.90	4.05	479	0.57
2q13dup(NPHP1)	hernia	335218	1654	48102	214	237.3	0.91	0.094	0.89	0.76	1.02	502	0.59
2q13_del	hernia	333605	41	47898	10	5.9	1.56	0.10	1.85	0.88	3.57	531	0.60
17p12_CMT1A_dup	hernia	333660	96	47908	20	13.8	1.37	0.12	1.50	0.90	2.39	582	0.63
15q11q13del_BP3-BP4	hernia	333575	11	47891	3	1.6	1.71	0.17	2.54	0.64	7.79	741	0.70
1q21.1_del	hernia	333653	89	47905	17	12.8	1.28	0.17	1.46	0.84	2.40	759	0.72
13q12.12_del	hernia	333641	77	47895	7	11.1	0.66	0.20	0.63	0.27	1.25	837	0.74
NRXN1_del	hernia	333697	133	47912	24	19.1	1.22	0.25	1.31	0.82	1.99	985	0.78
22q11.2_del	hernia	333572	8	47890	2	1.1	1.59	0.29	2.32	0.42	8.91	1141	0.81
15q13.3_dup	hernia	333763	199	47913	25	28.6	0.89	0.33	0.82	0.53	1.22	1259	0.82
13q12del_CRYL1	hernia	333890	326	47925	37	46.8	0.81	0.36	0.85	0.60	1.19	1335	0.84
16p13.11_dup	hernia	334241	677	47994	106	97.2	1.08	0.36	1.10	0.89	1.35	1340	0.84
3q29_del	hernia	333573	9	47888	0	1.3	0.00	0.38	0.34	0.00	2.75	1417	0.85
2q11.2_del	hernia	333588	24	47893	5	3.4	1.37	0.39	1.54	0.54	3.71	1439	0.85
15q24_dup	hernia	333571	7	47889	1	1.0	1.00	0.41	2.26	0.24	10.51	1491	0.86
17q11.2_del_NF1	hernia	333571	7	47890	2	1.0	1.77	0.44	1.82	0.33	7.11	1597	0.87
16p12.1_dup	hernia	333730	166	47914	26	23.8	1.08	0.51	1.15	0.74	1.72	1780	0.89
2q13del(NPHP1)	hernia	335612	2048	48170	282	293.9	0.96	0.52	0.96	0.84	1.09	1808	0.90
10q11.21q11.23_dup	hernia	333601	37	47891	3	5.3	0.60	0.54	0.72	0.19	1.90	1857	0.91
13q12.12_dup	hernia	333753	189	47911	23	27.1	0.86	0.58	0.89	0.56	1.34	1949	0.93
15q13.3_del	hernia	333595	31	47894	6	4.5	1.29	0.60	1.27	0.49	2.83	2011	0.94
17q12_dup	hernia	333648	84	47903	15	12.1	1.21	0.61	1.16	0.64	1.95	2052	0.94
8p23.1_dup	hernia	333569	5	47888	0	0.7	0.00	0.62	0.51	0.00	4.68	2076	0.94
10q23_dup	hernia	333570	6	47889	1	0.9	1.14	0.65	1.56	0.16	8.05	2156	0.95
16p11.2distal_dup	hernia	333675	111	47904	16	15.9	1.00	0.71	1.10	0.63	1.82	2318	0.96
2q21.1_dup	hernia	333613	49	47894	6	7.0	0.87	0.72	0.86	0.34	1.84	2338	0.96
15q11q13dup_BP3_BP5	hernia	333572	8	47889	1	1.1	0.89	0.72	1.40	0.15	6.50	2354	0.96
Potocki_Lupski	hernia	333569	5	47888	0	0.7	0.00	0.73	0.62	0.00	5.70	2377	0.96
22q11.2distal_dup	hernia	333575	11	47889	1	1.6	0.66	0.75	0.76	0.08	3.25	2428	0.97
10q11.21q11.23_del	hernia	333614	50	47894	6	7.2	0.85	0.75	0.88	0.35	1.87	2429	0.97
1q21.1_dup	hernia	333710	146	47910	22	21.0	1.04	0.77	1.07	0.67	1.65	2484	0.97
15q11.2_dup	hernia	335245	1681	48124	236	241.3	0.98	0.79	0.98	0.85	1.12	2547	0.97
16p11.2distal_del	hernia	333612	48	47894	6	6.9	0.89	0.80	0.90	0.36	1.93	2592	0.97
WBS_dup	hernia	333576	12	47889	1	1.7	0.61	0.81	0.81	0.09	3.48	2607	0.97
13q12dup_CRYL1	hernia	333573	9	47889	1	1.3	0.80	0.81	1.24	0.13	5.57	2617	0.97
15q11.2_del	hernia	334935	1371	48087	199	196.8	1.01	0.82	0.98	0.84	1.14	2652	0.97
22q11.2distal_del	hernia	333569	5	47888	0	0.7	0.00	0.83	0.73	0.01	6.80	2667	0.97
2q11.2_dup	hernia	333587	23	47891	3	3.3	0.92	0.83	1.14	0.30	3.15	2668	0.97
3q29_dup	hernia	333569	5	47888	0	0.72	0.00	0.86	0.78	0.01	7.00	2764	0.97
PWS_dup	hernia	333579	15	47889	1	2.2	0.50	0.92	0.91	0.10	3.84	2916	0.98
16p13.11_del	hernia	333674	110	47902	14	15.8	0.90	0.92	0.97	0.53	1.64	2927	0.98
15q13.3dup(CHRNA7)	hernia	336058	2494	48237	349	358.0	0.98	0.95	1.00	0.89	1.12	3008	0.99
15q13.3del(CHRNA7)	hernia	333573	9	47889	1	1.3	0.80	0.98	1.02	0.11	4.56	3089	1.00
TAR_del	hernia	333625	61	47896	8	8.8	0.92	0.99	1.00	0.45	1.96	3112	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2_del	high_cholesterol	314948	76	66607	27	16.1	1.50	0.0058	2.00	1.23	3.15	85	0.21
16p12.1_dup	high_cholesterol	315044	172	66600	20	36.4	0.60	0.0083	0.55	0.33	0.86	104	0.25
1q21.1_dup	high_cholesterol	314999	127	66621	41	26.9	1.40	0.013	1.63	1.11	2.34	142	0.29
15q11q13del_BP3-BP4	high_cholesterol	314881	9	66585	5	1.9	2.05	0.020	4.03	1.27	11.67	180	0.35
16p12.1_del	high_cholesterol	315054	182	66633	53	38.5	1.29	0.022	1.47	1.06	2.02	200	0.34
13q12.12_del	high_cholesterol	314933	61	66603	23	12.9	1.57	0.031	1.78	1.06	2.92	246	0.39
3q29_del	high_cholesterol	314877	5	66584	4	1.1	2.55	0.036	4.41	1.11	16.54	271	0.41
15q11.2_dup	high_cholesterol	316423	1551	66946	366	328.1	1.09	0.038	1.14	1.01	1.28	279	0.43
15q13.3_del	high_cholesterol	314897	25	66592	12	5.3	1.86	0.042	2.20	1.03	4.48	297	0.44
13q12.12_dup	high_cholesterol	315058	186	66606	26	39.3	0.70	0.065	0.68	0.44	1.02	379	0.54
15q13.3dup(CHRNA7)	high_cholesterol	317265	2393	67030	450	505.6	0.91	0.069	0.91	0.82	1.01	397	0.55
22q11.2distal_dup	high_cholesterol	314879	7	66585	5	1.5	2.39	0.078	2.91	0.88	9.03	442	0.55
17q11.2_del_NF1	high_cholesterol	314881	9	66580	0	1.9	0.00	0.098	0.16	0.00	1.30	521	0.59
15q24_dup	high_cholesterol	314878	6	66582	2	1.3	1.43	0.12	3.85	0.67	16.25	587	0.63
16p11.2distal_del	high_cholesterol	314913	41	66593	13	8.7	1.38	0.15	1.64	0.83	3.05	683	0.68
17q12_del	high_cholesterol	314876	4	66583	3	0.8	2.46	0.15	3.18	0.64	15.41	690	0.68
1q21.1_del	high_cholesterol	314966	94	66592	12	19.9	0.65	0.15	0.65	0.34	1.16	692	0.68
17q12_dup	high_cholesterol	314957	85	66594	14	18.0	0.81	0.21	0.70	0.38	1.21	869	0.75
15q13.3del(CHRNA7)	high_cholesterol	314879	7	66583	3	1.5	1.72	0.22	2.41	0.56	8.78	904	0.76
10q11.21q11.23_dup	high_cholesterol	314904	32	66588	8	6.8	1.15	0.23	1.66	0.70	3.54	938	0.78
15q13.3_dup	high_cholesterol	315061	189	66615	35	40.0	0.90	0.27	0.81	0.55	1.17	1068	0.80
16p11.2_dup	high_cholesterol	314984	112	66599	19	23.7	0.83	0.29	0.77	0.45	1.24	1139	0.81
2q21.1_del	high_cholesterol	314907	35	66585	5	7.4	0.72	0.31	0.63	0.22	1.48	1184	0.81
10q23_dup	high_cholesterol	314877	5	66582	2	1.1	1.64	0.31	2.47	0.38	12.68	1206	0.81
2q11.2_dup	high_cholesterol	314896	24	66582	2	5.1	0.44	0.32	0.53	0.11	1.71	1230	0.82
13q12del_CRYL1	high_cholesterol	315184	312	66631	51	66.0	0.80	0.33	0.86	0.63	1.16	1251	0.82
7q11.23_dup_distal	high_cholesterol	314890	18	66585	5	3.8	1.25	0.38	1.60	0.53	4.10	1391	0.85
8p23.1_dup	high_cholesterol	314877	5	66580	0	1.1	0.00	0.39	0.33	0.00	3.05	1425	0.85
TAR_dup	high_cholesterol	315204	332	66656	76	70.2	1.07	0.43	1.11	0.85	1.43	1548	0.87
16p11.2distal_dup	high_cholesterol	314975	103	66604	24	21.8	1.08	0.44	1.21	0.74	1.90	1592	0.87
10q11.21q11.23_del	high_cholesterol	314916	44	66592	12	9.3	1.23	0.45	1.30	0.64	2.45	1605	0.87
22q11.2_del	high_cholesterol	314880	8	66582	2	1.7	1.15	0.46	1.84	0.32	7.57	1627	0.88
2q11.2_del	high_cholesterol	314895	23	66586	6	4.9	1.19	0.47	1.42	0.52	3.38	1666	0.88
22q11.2distal_del	high_cholesterol	314876	4	66581	1	0.8	1.15	0.50	2.06	0.19	12.98	1746	0.89
13q12dup_CRYL1	high_cholesterol	314880	8	66582	2	1.7	1.15	0.52	1.67	0.30	6.47	1802	0.90
15q11.2_del	high_cholesterol	316170	1298	66852	272	274.5	0.99	0.52	0.96	0.83	1.09	1804	0.90
2q13_dup	high_cholesterol	314929	57	66594	14	12.1	1.13	0.53	1.22	0.64	2.19	1837	0.90
TAR_del	high_cholesterol	314928	56	66593	13	11.8	1.08	0.56	1.21	0.62	2.21	1923	0.92
17p12_CMT1A_dup	high_cholesterol	314966	94	66602	22	19.9	1.09	0.60	1.14	0.69	1.81	2020	0.94
16p13.11_del	high_cholesterol	314974	102	66602	22	21.6	1.02	0.64	1.12	0.68	1.78	2124	0.95
Potocki_Lupski	high_cholesterol	314876	4	66581	1	0.8	1.15	0.64	1.61	0.15	9.44	2128	0.95
PWS_dup	high_cholesterol	314887	15	66581	1	3.2	0.36	0.68	0.70	0.07	3.15	2228	0.95
3q29_dup	high_cholesterol	314877	5	66580	0	1.06	0.00	0.77	0.66	0.00	6.30	2496	0.97
17p12_HNPP_del	high_cholesterol	315050	178	66621	41	37.6	1.07	0.80	1.05	0.73	1.48	2575	0.97
2q13del(NPHP1)	high_cholesterol	316792	1920	66990	410	406.0	1.01	0.84	1.01	0.90	1.13	2688	0.97
NRXN1_del	high_cholesterol	315002	130	66607	27	27.5	0.99	0.85	1.04	0.67	1.58	2734	0.97
2q13dup(NPHP1)	high_cholesterol	316411	1539	66909	329	325.4	1.01	0.86	0.99	0.87	1.12	2769	0.97
2q21.1_dup	high_cholesterol	314917	45	66590	10	9.5	1.04	0.87	1.06	0.50	2.06	2798	0.98
16p13.11_dup	high_cholesterol	315520	648	66715	135	137.0	0.99	0.95	1.01	0.83	1.22	3012	0.99
22q11.2_dup	high_cholesterol	315092	220	66626	46	46.5	0.99	0.96	1.01	0.71	1.40	3044	0.99
2q13_del	high_cholesterol	314915	43	66588	8	9.1	0.90	0.97	1.01	0.44	2.09	3071	0.99
WBS_dup	high_cholesterol	314883	11	66582	2	2.3	0.88	0.98	1.02	0.19	3.77	3078	0.99
15q11q13dup_BP3_BP4	high_cholesterol	314914	42	66588	8	8.9	0.92	0.99	1.00	0.44	2.07	3110	1.00
15q11q13dup_BP3_BP5	high_cholesterol	314880	8	66581	1	1.7	0.64	1.00	1.00	0.10	4.94	3128	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p12.1_del	hypertension	260945	124	120742	111	57.4	1.49	8.64E-08	2.11	1.61	2.76	5	5.41E-05*
16p11.2_del	hypertension	260873	52	120682	51	24.1	1.57	9.44E-06	2.55	1.69	3.83	12	0.0025*
16p13.11_dup	hypertension	261304	483	120931	300	223.5	1.21	2.05E-05	1.39	1.20	1.62	15	0.0043*
13q12.12_del	hypertension	260867	46	120669	38	21.3	1.43	0.014	1.77	1.12	2.77	152	0.29
22q11.2_del	hypertension	260825	4	120637	6	1.9	1.90	0.020	4.61	1.28	18.00	176	0.35
1q21.1_dup	hypertension	260923	102	120697	66	47.2	1.24	0.023	1.47	1.06	2.02	205	0.35
15q13.3_del	hypertension	260840	19	120649	18	8.8	1.54	0.050	1.98	1.00	3.92	328	0.47
TAR_del	hypertension	260862	41	120659	28	19.0	1.28	0.053	1.67	0.99	2.75	339	0.49
10q11.21q11.23_dup	hypertension	260845	24	120647	16	11.1	1.26	0.054	1.94	0.99	3.71	348	0.49
2q13_del	hypertension	260851	30	120652	21	13.9	1.30	0.054	1.79	0.99	3.20	349	0.49
2q13dup(NPHP1)	hypertension	262132	1311	121188	557	606.1	0.94	0.055	0.90	0.81	1.00	352	0.49
TAR_dup	hypertension	261085	264	120775	144	122.1	1.12	0.067	1.22	0.99	1.51	385	0.54
16p11.2distal_dup	hypertension	260902	81	120677	46	37.5	1.15	0.081	1.41	0.96	2.05	455	0.56
22q11.2_dup	hypertension	260992	171	120726	95	79.1	1.13	0.086	1.26	0.97	1.64	474	0.57
17q11.2_del_NF1	hypertension	260829	8	120632	1	3.7	0.35	0.094	0.26	0.03	1.22	501	0.59
22q11.2distal_del	hypertension	260823	2	120634	3	0.9	1.90	0.096	4.32	0.76	27.80	513	0.59
8p23.1_dup	hypertension	260826	5	120631	0	2.3	0.00	0.097	0.14	0.00	1.32	514	0.59
16p11.2distal_del	hypertension	260853	32	120653	22	14.8	1.29	0.11	1.59	0.90	2.78	560	0.63
17q12_dup	hypertension	260881	60	120670	39	27.8	1.25	0.15	1.37	0.89	2.09	684	0.68
13q12.12_dup	hypertension	260977	156	120687	56	72.1	0.84	0.15	0.80	0.58	1.09	695	0.68
17p12_HNPP_del	hypertension	260960	139	120711	80	64.3	1.16	0.18	1.22	0.91	1.62	793	0.73
2q21.1_del	hypertension	260844	23	120648	17	10.6	1.34	0.20	1.55	0.79	2.96	827	0.74
NRXN1_del	hypertension	260923	102	120686	55	47.2	1.11	0.21	1.25	0.88	1.75	877	0.75
15q11q13dup_BP3_BP5	hypertension	260826	5	120635	4	2.3	1.41	0.23	2.31	0.58	8.86	923	0.77
15q11.2_dup	hypertension	262111	1290	121258	627	596.8	1.03	0.24	1.06	0.96	1.17	960	0.78
15q13.3dup(CHRNA7)	hypertension	262751	1930	121544	913	892.8	1.02	0.25	1.05	0.97	1.14	1002	0.78
2q11.2_del	hypertension	260844	23	120637	6	10.6	0.65	0.26	0.61	0.23	1.42	1026	0.79
15q13.3_dup	hypertension	260965	144	120711	80	66.6	1.13	0.30	1.16	0.87	1.55	1152	0.81
3q29_dup	hypertension	260824	3	120633	2	1.39	1.26	0.31	2.45	0.39	13.56	1205	0.81
16p12.1_dup	hypertension	260960	139	120684	53	64.3	0.87	0.32	0.85	0.60	1.17	1219	0.82
15q11q13del_BP3-BP4	hypertension	260833	12	120633	2	5.5	0.45	0.32	0.52	0.10	1.78	1232	0.82
16p11.2_dup	hypertension	260915	94	120668	37	43.5	0.89	0.33	0.82	0.55	1.21	1254	0.82
15q11q13dup_BP3_BP4	hypertension	260853	32	120649	18	14.8	1.14	0.34	1.35	0.73	2.43	1279	0.82
22q11.2distal_dup	hypertension	260827	6	120637	6	2.8	1.58	0.35	1.70	0.55	5.32	1318	0.83
10q23_dup	hypertension	260825	4	120634	3	1.9	1.36	0.38	2.01	0.40	9.43	1411	0.85
3q29_del	hypertension	260826	5	120635	4	2.3	1.41	0.39	1.78	0.46	6.54	1435	0.85
2q13del(NPHP1)	hypertension	262396	1575	121386	755	728.6	1.02	0.40	1.04	0.95	1.14	1483	0.85
13q12dup_CRYL1	hypertension	260827	6	120635	4	2.8	1.26	0.42	1.68	0.45	5.76	1529	0.87
17p12_CMT1A_dup	hypertension	260904	83	120664	33	38.4	0.90	0.48	0.86	0.56	1.30	1690	0.88
15q13.3del(CHRNA7)	hypertension	260827	6	120635	4	2.8	1.26	0.50	1.55	0.41	5.43	1760	0.89
2q11.2_dup	hypertension	260841	20	120637	6	9.2	0.73	0.52	0.75	0.28	1.77	1820	0.90
2q13_dup	hypertension	260868	47	120655	24	21.7	1.07	0.56	1.17	0.69	1.94	1918	0.91
2q21.1_dup	hypertension	260860	39	120647	16	18.0	0.92	0.64	0.87	0.47	1.55	2123	0.95
15q11.2_del	hypertension	261880	1059	121142	511	489.9	1.03	0.65	1.03	0.92	1.15	2137	0.95
Potocki_Lupski	hypertension	260824	3	120633	2	1.4	1.26	0.65	1.49	0.24	8.03	2144	0.95
15q24_dup	hypertension	260827	6	120633	2	2.8	0.79	0.66	1.41	0.25	5.80	2183	0.95
16p13.11_del	hypertension	260906	85	120670	39	39.3	0.99	0.74	1.07	0.71	1.58	2411	0.96
PWS_dup	hypertension	260834	13	120634	3	6.0	0.59	0.79	0.85	0.21	2.69	2562	0.97
17q12_del	hypertension	260826	5	120633	2	2.3	0.90	0.80	0.82	0.14	3.72	2586	0.97
10q11.21q11.23_del	hypertension	260858	37	120650	19	17.1	1.07	0.81	1.07	0.59	1.88	2614	0.97
7q11.23_dup_distal	hypertension	260837	16	120638	7	7.4	0.96	0.92	1.04	0.40	2.49	2939	0.99
13q12del_CRYL1	hypertension	261077	256	120738	107	118.4	0.93	0.94	0.99	0.78	1.25	2979	0.99
WBS_dup	hypertension	260830	9	120635	4	4.2	0.97	0.94	1.05	0.29	3.30	2988	0.99
1q21.1_del	hypertension	260895	74	120663	32	34.2	0.95	0.96	1.01	0.65	1.54	3029	0.99



CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
3q29_dup	inflammatory_bowel_disease	362864	2	18593	3	0.10	12.31	0.00013	35.45	6.68	217.84	20	0.021*
2q11.2_dup	inflammatory_bowel_disease	362883	21	18595	5	1.1	3.95	0.0040	5.16	1.81	12.37	73	0.17
13q12dup_CRYL1	inflammatory_bowel_disease	362869	7	18593	3	0.4	6.16	0.0058	8.37	2.02	28.32	86	0.21
16p11.2distal_dup	inflammatory_bowel_disease	362976	114	18603	13	5.8	2.10	0.0091	2.31	1.25	3.93	113	0.25
15q13.3_del	inflammatory_bowel_disease	362894	32	18595	5	1.6	2.77	0.043	2.89	1.04	6.63	301	0.45
15q13.3del(CHRNA7)	inflammatory_bowel_disease	362870	8	18592	2	0.4	4.10	0.045	5.54	1.04	20.23	313	0.46
TAR_del	inflammatory_bowel_disease	362931	69	18590	0	3.5	0.00	0.048	0.14	0.00	0.99	323	0.47
16p12.1_del	inflammatory_bowel_disease	363091	229	18596	6	11.7	0.52	0.081	0.53	0.22	1.07	456	0.56
1q21.1_del	inflammatory_bowel_disease	362959	97	18599	9	5.0	1.74	0.089	1.88	0.90	3.48	487	0.57
15q11.2_del	inflammatory_bowel_disease	364370	1508	18652	62	77.2	0.81	0.092	0.81	0.62	1.03	496	0.58
8p23.1_dup	inflammatory_bowel_disease	362866	4	18591	1	0.2	4.10	0.097	6.47	0.65	35.15	520	0.59
1q21.1_dup	inflammatory_bowel_disease	363017	155	18603	13	7.9	1.59	0.11	1.62	0.89	2.73	559	0.63
PWS_dup	inflammatory_bowel_disease	362876	14	18592	2	0.7	2.56	0.12	3.48	0.68	11.43	585	0.63
22q11.2distal_del	inflammatory_bowel_disease	362866	4	18591	1	0.2	4.10	0.12	5.71	0.56	31.61	598	0.63
15q11q13dup_BP3_BP4	inflammatory_bowel_disease	362912	50	18590	0	2.6	0.00	0.12	0.20	0.00	1.38	616	0.64
15q13.3dup(CHRNA7)	inflammatory_bowel_disease	365551	2689	18744	154	137.9	1.11	0.15	1.13	0.96	1.32	698	0.68
17q12_del	inflammatory_bowel_disease	362868	6	18591	1	0.3	2.93	0.18	4.12	0.43	19.70	780	0.73
15q24_dup	inflammatory_bowel_disease	362869	7	18591	1	0.4	2.56	0.19	3.98	0.42	18.32	803	0.73
13q12.12_dup	inflammatory_bowel_disease	363060	198	18604	14	10.1	1.36	0.21	1.43	0.80	2.35	889	0.76
10q11.21q11.23_dup	inflammatory_bowel_disease	362902	40	18590	0	2.0	0.00	0.23	0.26	0.00	1.82	925	0.77
15q11q13dup_BP3_BP5	inflammatory_bowel_disease	362870	8	18591	1	0.4	2.28	0.24	3.31	0.35	14.81	970	0.78
3q29_del	inflammatory_bowel_disease	362870	8	18591	1	0.4	2.28	0.25	3.26	0.35	14.57	986	0.78
13q12del_CRYL1	inflammatory_bowel_disease	363204	342	18611	21	17.5	1.19	0.27	1.29	0.81	1.94	1072	0.80
17p12_CMT1A_dup	inflammatory_bowel_disease	362970	108	18598	8	5.5	1.42	0.28	1.50	0.69	2.84	1107	0.80
2q13_dup	inflammatory_bowel_disease	362928	66	18595	5	3.4	1.45	0.29	1.66	0.61	3.62	1123	0.81
16p13.11_dup	inflammatory_bowel_disease	363613	751	18622	32	38.5	0.84	0.30	0.83	0.57	1.16	1142	0.81
2q21.1_dup	inflammatory_bowel_disease	362913	51	18594	4	2.6	1.49	0.31	1.72	0.56	4.06	1178	0.81
2q11.2_del	inflammatory_bowel_disease	362891	29	18590	0	1.5	0.00	0.33	0.32	0.00	2.29	1265	0.82
2q21.1_del	inflammatory_bowel_disease	362899	37	18593	3	1.9	1.54	0.35	1.76	0.48	4.61	1313	0.83
16p11.2distal_del	inflammatory_bowel_disease	362915	53	18591	1	2.7	0.38	0.39	0.53	0.06	1.94	1453	0.85
15q11q13del_BP3-BP4	inflammatory_bowel_disease	362875	13	18591	1	0.7	1.47	0.39	2.27	0.25	9.34	1457	0.85
WBS_dup	inflammatory_bowel_disease	362874	12	18591	1	0.6	1.58	0.41	2.19	0.24	9.18	1500	0.86
17p12_HNPP_del	inflammatory_bowel_disease	363073	211	18598	8	10.8	0.75	0.47	0.78	0.36	1.46	1680	0.88
2q13dup(NPHP1)	inflammatory_bowel_disease	364632	1770	18688	98	90.7	1.08	0.47	1.08	0.87	1.32	1682	0.88
16p12.1_dup	inflammatory_bowel_disease	363047	185	18597	7	9.5	0.75	0.49	0.78	0.34	1.51	1745	0.89
15q13.3_dup	inflammatory_bowel_disease	363073	211	18603	13	10.8	1.19	0.50	1.22	0.67	2.03	1757	0.89
13q12.12_del	inflammatory_bowel_disease	362941	79	18595	5	4.0	1.22	0.55	1.32	0.49	2.86	1879	0.91
2q13_del	inflammatory_bowel_disease	362910	48	18593	3	2.5	1.21	0.55	1.42	0.39	3.65	1888	0.91
TAR_dup	inflammatory_bowel_disease	363249	387	18611	21	19.8	1.06	0.70	1.09	0.68	1.64	2300	0.96
7q11.23_dup_distal	inflammatory_bowel_disease	362884	22	18591	1	1.1	0.89	0.73	1.36	0.15	5.28	2372	0.96
NRXN1_del	inflammatory_bowel_disease	363011	149	18598	8	7.6	1.05	0.74	1.13	0.52	2.11	2412	0.96
10q11.21q11.23_del	inflammatory_bowel_disease	362916	54	18592	2	2.8	0.73	0.74	0.81	0.17	2.38	2415	0.97
22q11.2distal_dup	inflammatory_bowel_disease	362874	12	18590	0	0.6	0.00	0.76	0.66	0.01	5.03	2449	0.97
Potocki_Lupski	inflammatory_bowel_disease	362867	5	18590	0	0.3	0.00	0.76	1.64	0.01	14.48	2452	0.97
16p11.2_dup	inflammatory_bowel_disease	362986	124	18597	7	6.4	1.10	0.76	1.13	0.49	2.20	2453	0.96
16p13.11_del	inflammatory_bowel_disease	362981	119	18595	5	6.1	0.83	0.77	0.88	0.33	1.89	2497	0.97
17q12_dup	inflammatory_bowel_disease	362956	94	18595	5	4.8	1.04	0.77	1.14	0.42	2.45	2504	0.97
15q11.2_dup	inflammatory_bowel_disease	364681	1819	18688	98	93.2	1.05	0.77	1.03	0.83	1.26	2511	0.97
22q11.2_dup	inflammatory_bowel_disease	363116	254	18602	12	13.0	0.93	0.80	0.93	0.50	1.57	2588	0.97
10q23_dup	inflammatory_bowel_disease	362869	7	18590	0	0.4	0.00	0.82	1.42	0.01	11.79	2650	0.97
2q13del(NPHP1)	inflammatory_bowel_disease	365077	2215	18705	115	113.5	1.01	0.85	1.02	0.84	1.22	2742	0.97
22q11.2_del	inflammatory_bowel_disease	362872	10	18590	0	0.5	0.00	0.93	0.88	0.01	6.92	2956	0.99
16p11.2_del	inflammatory_bowel_disease	362960	98	18595	5	5.0	1.00	0.95	0.97	0.36	2.09	2999	0.99
17q11.2_del_NF1	inflammatory_bowel_disease	362871	9	18590	0	0.5	0.00	0.99	0.98	0.01	7.71	3098	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2_dup	irritable_bowel_syndrome	368567	118	13016	13	4.2	2.91	0.00036	3.33	1.80	5.67	30	0.037*
PWS_dup	irritable_bowel_syndrome	368462	13	13006	3	0.5	5.50	0.011	6.55	1.66	19.70	128	0.26
3q29_del	irritable_bowel_syndrome	368456	7	13005	2	0.2	6.52	0.012	10.06	1.83	39.47	137	0.28
1q21.1_dup	irritable_bowel_syndrome	368605	156	13015	12	5.5	2.10	0.017	2.20	1.17	3.77	167	0.31
15q11q13del_BP3-BP4	irritable_bowel_syndrome	368461	12	13005	2	0.4	4.19	0.035	6.03	1.16	20.68	267	0.42
8p23.1_dup	irritable_bowel_syndrome	368453	4	13004	1	0.1	5.87	0.041	11.60	1.13	66.28	296	0.44
22q11.2_dup	irritable_bowel_syndrome	368700	251	13018	15	8.9	1.65	0.045	1.77	1.01	2.87	311	0.45
16p12.1_dup	irritable_bowel_syndrome	368630	181	13014	11	6.4	1.68	0.093	1.74	0.90	3.03	499	0.59
15q13.3_del	irritable_bowel_syndrome	368483	34	13006	3	1.2	2.38	0.11	2.81	0.76	7.49	553	0.62
10q11.21q11.23_del	irritable_bowel_syndrome	368501	52	13007	4	1.8	2.10	0.14	2.27	0.74	5.37	661	0.65
15q11.2_del	irritable_bowel_syndrome	369977	1528	13045	42	53.9	0.78	0.14	0.80	0.58	1.07	677	0.67
15q24_dup	irritable_bowel_syndrome	368456	7	13004	1	0.2	3.67	0.15	4.68	0.49	21.82	687	0.68
2q11.2_del	irritable_bowel_syndrome	368476	27	13005	2	1.0	2.02	0.18	2.74	0.55	8.45	791	0.73
16p11.2_del	irritable_bowel_syndrome	368551	102	13004	1	3.6	0.28	0.20	0.41	0.05	1.47	853	0.74
17q12_dup	irritable_bowel_syndrome	368543	94	13008	5	3.3	1.48	0.21	1.84	0.68	3.98	862	0.75
17p12_HNPP_del	irritable_bowel_syndrome	368664	215	13007	4	7.6	0.54	0.24	0.60	0.20	1.35	961	0.78
TAR_dup	irritable_bowel_syndrome	368840	391	13020	17	13.8	1.22	0.28	1.32	0.78	2.07	1093	0.80
16p11.2distal_dup	irritable_bowel_syndrome	368570	121	13009	6	4.3	1.39	0.35	1.49	0.61	3.04	1323	0.83
2q21.1_del	irritable_bowel_syndrome	368489	40	13003	0	1.4	0.00	0.37	0.35	0.00	2.45	1378	0.85
2q13del(NPHP1)	irritable_bowel_syndrome	370692	2243	13090	87	79.2	1.10	0.42	1.09	0.88	1.35	1503	0.87
13q12.12_dup	irritable_bowel_syndrome	368652	203	13012	9	7.2	1.25	0.46	1.29	0.63	2.34	1643	0.88
15q11q13dup_BP3_BP4	irritable_bowel_syndrome	368497	48	13005	2	1.7	1.17	0.49	1.63	0.33	4.81	1723	0.88
7q11.23_dup_distal	irritable_bowel_syndrome	368471	22	13004	1	0.8	1.28	0.49	1.89	0.21	7.38	1741	0.89
3q29_dup	irritable_bowel_syndrome	368454	5	13003	0	0.18	0.00	0.50	3.29	0.02	31.43	1749	0.89
2q13_dup	irritable_bowel_syndrome	368519	70	13004	1	2.5	0.41	0.53	0.62	0.07	2.27	1846	0.90
2q11.2_dup	irritable_bowel_syndrome	368474	25	13004	1	0.9	1.13	0.56	1.70	0.19	6.58	1916	0.91
15q13.3_dup	irritable_bowel_syndrome	368665	216	13011	8	7.6	1.05	0.56	1.23	0.57	2.30	1924	0.92
Potocki Lupski	irritable_bowel_syndrome	368454	5	13003	0	0.2	0.00	0.59	2.43	0.02	22.23	1999	0.93
10q23_dup	irritable_bowel_syndrome	368456	7	13003	0	0.2	0.00	0.62	2.25	0.02	19.38	2066	0.94
17q12_del	irritable_bowel_syndrome	368456	7	13003	0	0.2	0.00	0.65	2.08	0.02	17.69	2159	0.95
22q11.2distal_del	irritable_bowel_syndrome	368454	5	13003	0	0.2	0.00	0.66	2.03	0.02	18.80	2189	0.95
2q13dup(NPHP1)	irritable_bowel_syndrome	370251	1802	13069	66	63.6	1.04	0.67	1.06	0.82	1.34	2199	0.95
16p13.11_del	irritable_bowel_syndrome	368568	119	13008	5	4.2	1.18	0.68	1.20	0.45	2.58	2232	0.95
13q12del_CRYL1	irritable_bowel_syndrome	368799	350	13016	13	12.4	1.05	0.72	1.11	0.61	1.83	2337	0.96
1q21.1_del	irritable_bowel_syndrome	368551	102	13007	4	3.6	1.11	0.72	1.19	0.39	2.74	2361	0.96
16p13.11_dup	irritable_bowel_syndrome	369204	755	13031	28	26.6	1.05	0.74	1.07	0.72	1.52	2395	0.96
17q11.2_del_NF1	irritable_bowel_syndrome	368458	9	13003	0	0.3	0.00	0.76	1.60	0.01	12.99	2471	0.97
16p12.1_del	irritable_bowel_syndrome	368677	228	13010	7	8.0	0.87	0.77	0.90	0.40	1.74	2509	0.97
16p11.2distal_del	irritable_bowel_syndrome	368502	53	13004	1	1.9	0.54	0.78	0.80	0.09	2.97	2530	0.97
15q13.3del(CHRNA7)	irritable_bowel_syndrome	368459	10	13003	0	0.4	0.00	0.78	1.53	0.01	12.27	2537	0.97
17p12_CMT1A_dup	irritable_bowel_syndrome	368562	113	13006	3	4.0	0.76	0.80	0.87	0.24	2.19	2585	0.97
13q12.12_del	irritable_bowel_syndrome	368531	82	13005	2	2.9	0.70	0.81	0.86	0.18	2.48	2603	0.97
15q11q13dup_BP3_BP5	irritable_bowel_syndrome	368458	9	13003	0	0.3	0.00	0.83	1.38	0.01	11.11	2676	0.97
10q11.21q11.23_dup	irritable_bowel_syndrome	368488	39	13004	1	1.4	0.73	0.84	1.19	0.13	4.48	2697	0.97
2q21.1_dup	irritable_bowel_syndrome	368503	54	13004	1	1.9	0.53	0.84	0.85	0.10	3.14	2700	0.97
22q11.2_del	irritable_bowel_syndrome	368459	10	13003	0	0.4	0.00	0.84	1.36	0.01	10.93	2701	0.97
2q13_del	irritable_bowel_syndrome	368499	50	13004	1	1.8	0.58	0.87	0.88	0.10	3.25	2789	0.98
13q12dup_CRYL1	irritable_bowel_syndrome	368459	10	13003	0	0.4	0.00	0.92	1.17	0.01	9.25	2914	0.98
15q11.2_dup	irritable_bowel_syndrome	370301	1852	13068	65	65.4	0.99	0.93	0.99	0.76	1.26	2947	0.98
TAR_del	irritable_bowel_syndrome	368516	67	13005	2	2.4	0.85	0.94	1.05	0.22	3.06	2964	0.99
NRXN1_del	irritable_bowel_syndrome	368601	152	13008	5	5.4	0.93	0.94	1.03	0.39	2.20	2990	0.99
22q11.2distal_dup	irritable_bowel_syndrome	368461	12	13003	0	0.4	0.00	0.95	1.10	0.01	8.56	3005	0.99
15q13.3dup(CHRNA7)	irritable_bowel_syndrome	371196	2747	13099	96	96.9	0.99	0.98	1.00	0.81	1.22	3095	1.00
WBS_dup	irritable_bowel_syndrome	368462	13	13003	0	0.5	0.00	1.00	1.00	0.01	7.70	3127	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p12.1_del	ischaemic_heart_disease_not_MI	365344	215	16343	20	9.6	1.99	0.0023	2.22	1.35	3.45	61	0.12
16p11.2distal_dup	ischaemic_heart_disease_not_MI	365245	116	16334	11	5.2	2.02	0.0085	2.55	1.30	4.56	107	0.25
22q11.2_dup	ischaemic_heart_disease_not_MI	365375	246	16343	20	11.0	1.76	0.0087	1.96	1.20	3.03	109	0.25
15q11q13dup_BP3_BP5	ischaemic_heart_disease_not_MI	365136	7	16325	2	0.3	5.19	0.013	10.30	1.80	43.33	141	0.28
15q13.3del(CHRNA7)	ischaemic_heart_disease_not_MI	365137	8	16325	2	0.4	4.67	0.027	7.22	1.31	28.17	228	0.37
2q11.2_dup	ischaemic_heart_disease_not_MI	365152	23	16326	3	1.0	2.70	0.043	4.05	1.05	11.48	300	0.45
15q11.2_dup	ischaemic_heart_disease_not_MI	366947	1818	16422	99	81.4	1.21	0.044	1.24	1.01	1.52	306	0.45
16p11.2_del	ischaemic_heart_disease_not_MI	365224	95	16331	8	4.2	1.82	0.071	2.08	0.93	4.06	413	0.54
15q11.2_del	ischaemic_heart_disease_not_MI	366644	1515	16378	55	67.7	0.82	0.075	0.79	0.59	1.02	425	0.55
2q13del(NPHP1)	ischaemic_heart_disease_not_MI	367342	2213	16440	117	99.0	1.17	0.075	1.19	0.98	1.43	427	0.55
2q13_dup	ischaemic_heart_disease_not_MI	365194	65	16329	6	2.9	1.97	0.078	2.28	0.90	4.86	440	0.55
2q21.1_dup	ischaemic_heart_disease_not_MI	365184	55	16323	0	2.5	0.00	0.12	0.19	0.00	1.34	577	0.63
15q13.3_dup	ischaemic_heart_disease_not_MI	365338	209	16338	15	9.3	1.56	0.12	1.55	0.88	2.55	605	0.63
1q21.1_dup	ischaemic_heart_disease_not_MI	365286	157	16334	11	7.0	1.53	0.13	1.67	0.85	2.94	621	0.64
1q21.1_del	ischaemic_heart_disease_not_MI	365228	99	16330	7	4.4	1.54	0.14	1.86	0.80	3.72	663	0.65
22q11.2_del	ischaemic_heart_disease_not_MI	365138	9	16324	1	0.4	2.34	0.17	4.50	0.45	22.09	737	0.70
13q12dup_CRYL1	ischaemic_heart_disease_not_MI	365138	9	16324	1	0.4	2.34	0.18	4.21	0.44	19.50	764	0.72
16p11.2distal_del	ischaemic_heart_disease_not_MI	365179	50	16327	4	2.2	1.73	0.18	2.11	0.68	5.06	767	0.72
2q13dup(NPHP1)	ischaemic_heart_disease_not_MI	366928	1799	16392	69	80.4	0.86	0.18	0.85	0.66	1.07	768	0.72
17q12_del	ischaemic_heart_disease_not_MI	365135	6	16324	1	0.3	3.34	0.18	4.30	0.43	22.72	773	0.72
3q29_del	ischaemic_heart_disease_not_MI	365137	8	16324	1	0.4	2.60	0.19	4.08	0.42	19.47	797	0.73
15q11q13del_BP3-BP4	ischaemic_heart_disease_not_MI	365142	13	16324	1	0.6	1.67	0.20	3.69	0.40	15.45	851	0.74
15q13.3dup(CHRNA7)	ischaemic_heart_disease_not_MI	367841	2712	16454	131	121.3	1.08	0.22	1.12	0.93	1.33	895	0.76
13q12.12_del	ischaemic_heart_disease_not_MI	365207	78	16329	6	3.5	1.67	0.23	1.71	0.68	3.60	931	0.77
16p11.2_dup	ischaemic_heart_disease_not_MI	365257	128	16326	3	5.7	0.54	0.25	0.56	0.16	1.42	1003	0.78
10q11.21q11.23_del	ischaemic_heart_disease_not_MI	365181	52	16327	4	2.3	1.67	0.26	1.85	0.59	4.48	1027	0.79
NRXN1_del	ischaemic_heart_disease_not_MI	365277	148	16332	9	6.6	1.34	0.26	1.50	0.72	2.78	1030	0.79
16p13.11_del	ischaemic_heart_disease_not_MI	365246	117	16330	7	5.2	1.32	0.29	1.54	0.66	3.07	1122	0.81
2q21.1_del	ischaemic_heart_disease_not_MI	365166	37	16326	3	1.7	1.75	0.29	1.92	0.52	5.18	1129	0.81
WBS_dup	ischaemic_heart_disease_not_MI	365141	12	16324	1	0.5	1.80	0.31	2.78	0.29	12.35	1183	0.81
13q12del_CRYL1	ischaemic_heart_disease_not_MI	365474	345	16341	18	15.4	1.16	0.32	1.29	0.77	2.04	1226	0.81
TAR_del	ischaemic_heart_disease_not_MI	365197	68	16324	1	3.0	0.34	0.34	0.49	0.06	1.84	1301	0.83
15q11q13dup_BP3_BP4	ischaemic_heart_disease_not_MI	365176	47	16326	3	2.1	1.40	0.35	1.77	0.48	4.68	1315	0.83
22q11.2distal_dup	ischaemic_heart_disease_not_MI	365140	11	16324	1	0.5	1.95	0.38	2.34	0.25	10.23	1418	0.85
2q11.2_del	ischaemic_heart_disease_not_MI	365158	29	16323	0	1.3	0.00	0.42	0.38	0.00	2.76	1530	0.87
3q29_dup	ischaemic_heart_disease_not_MI	365134	5	16323	0	0.22	0.00	0.46	3.64	0.03	34.02	1649	0.88
16p12.1_dup	ischaemic_heart_disease_not_MI	365315	186	16329	6	8.3	0.73	0.55	0.79	0.32	1.61	1891	0.92
15q24_dup	ischaemic_heart_disease_not_MI	365137	8	16323	0	0.4	0.00	0.56	2.66	0.02	22.57	1920	0.92
22q11.2distal_del	ischaemic_heart_disease_not_MI	365134	5	16323	0	0.2	0.00	0.58	2.56	0.02	25.52	1955	0.93
7q11.23_dup_distal	ischaemic_heart_disease_not_MI	365152	23	16323	0	1.0	0.00	0.61	0.52	0.00	3.83	2033	0.94
16p13.11_dup	ischaemic_heart_disease_not_MI	365877	748	16358	35	33.4	1.04	0.64	1.09	0.76	1.50	2117	0.94
13q12.12_dup	ischaemic_heart_disease_not_MI	365334	205	16330	7	9.2	0.77	0.66	0.85	0.37	1.66	2187	0.95
Potocki Lupski	ischaemic_heart_disease_not_MI	365134	5	16323	0	0.2	0.00	0.69	1.95	0.01	18.88	2255	0.95
15q13.3_del	ischaemic_heart_disease_not_MI	365164	35	16325	2	1.6	1.26	0.69	1.32	0.27	4.08	2257	0.95
8p23.1_dup	ischaemic_heart_disease_not_MI	365134	5	16323	0	0.2	0.00	0.75	1.68	0.01	15.62	2421	0.97
2q13_del	ischaemic_heart_disease_not_MI	365178	49	16325	2	2.2	0.92	0.75	1.24	0.25	3.72	2448	0.97
17p12_CMT1A_dup	ischaemic_heart_disease_not_MI	365241	112	16327	4	5.0	0.81	0.81	0.89	0.29	2.07	2609	0.97
10q23_dup	ischaemic_heart_disease_not_MI	365136	7	16323	0	0.3	0.00	0.83	1.41	0.01	13.53	2664	0.97
TAR_dup	ischaemic_heart_disease_not_MI	365521	392	16339	16	17.5	0.92	0.85	0.95	0.56	1.52	2735	0.97
17p12_HNPP_del	ischaemic_heart_disease_not_MI	365339	210	16332	9	9.4	0.96	0.89	0.96	0.46	1.74	2844	0.98
17q11.2_del_NF1	ischaemic_heart_disease_not_MI	365138	9	16323	0	0.4	0.00	0.90	0.84	0.01	6.92	2881	0.98
17q12_dup	ischaemic_heart_disease_not_MI	365224	95	16327	4	4.2	0.94	0.91	0.94	0.31	2.20	2890	0.98
10q11.21q11.23_dup	ischaemic_heart_disease_not_MI	365168	39	16324	1	1.7	0.58	0.93	1.08	0.12	4.17	2950	0.98
PWS_dup	ischaemic_heart_disease_not_MI	365145	16	16323	0	0.7	0.00	0.98	1.04	0.01	8.50	3080	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
3q29_dup	low_WBC_count	378491	4	2966	1	0.03	25.73	0.0034	54.22	5.38	297.82	69	0.16
17q12_del	low_WBC_count	378493	6	2966	1	0.0	18.38	0.0072	32.49	3.38	154.82	99	0.23
16p13.11_del	low_WBC_count	378607	120	2969	4	0.9	4.15	0.0095	4.69	1.56	10.72	121	0.25
22q11.2_dup	low_WBC_count	378747	260	2971	6	2.0	2.90	0.011	3.33	1.37	6.68	129	0.27
22q11.2_del	low_WBC_count	378496	9	2966	1	0.1	12.87	0.011	23.63	2.53	103.05	132	0.27
13q12.12_del	low_WBC_count	378568	81	2968	3	0.6	4.59	0.014	5.40	1.50	13.62	153	0.29
13q12dup_CRYL1	low_WBC_count	378496	9	2966	1	0.1	12.87	0.016	19.00	2.03	82.75	163	0.31
2q13_del	low_WBC_count	378536	49	2967	2	0.4	5.05	0.021	7.00	1.44	20.54	188	0.35
16p11.2_del	low_WBC_count	378587	100	2968	3	0.8	3.75	0.021	4.74	1.32	11.90	196	0.34
13q12del_CRYL1	low_WBC_count	378845	358	2970	5	2.8	1.77	0.14	2.03	0.76	4.26	669	0.66
15q13.3dup(CHRNA7)	low_WBC_count	381302	2815	2993	28	22.1	1.27	0.19	1.29	0.87	1.84	813	0.74
2q21.1_dup	low_WBC_count	378541	54	2966	1	0.4	2.34	0.19	3.66	0.41	13.47	818	0.74
22q11.2distal_del	low_WBC_count	378492	5	2965	0	0.0	0.00	0.21	12.44	0.09	111.13	881	0.75
10q11.21q11.23_del	low_WBC_count	378542	55	2966	1	0.4	2.30	0.22	3.34	0.38	12.30	905	0.77
8p23.1_dup	low_WBC_count	378492	5	2965	0	0.0	0.00	0.22	11.42	0.09	101.25	910	0.77
Potocki_Lupski	low_WBC_count	378492	5	2965	0	0.0	0.00	0.22	11.36	0.09	100.84	913	0.77
10q23_dup	low_WBC_count	378494	7	2965	0	0.1	0.00	0.24	10.18	0.08	84.17	958	0.78
2q13del(NPHP1)	low_WBC_count	380804	2317	2978	13	18.1	0.72	0.26	0.74	0.41	1.22	1014	0.79
TAR_del	low_WBC_count	378555	68	2966	1	0.5	1.86	0.28	2.84	0.32	10.38	1086	0.80
15q24_dup	low_WBC_count	378495	8	2965	0	0.1	0.00	0.28	7.91	0.06	63.99	1088	0.80
2q13dup(NPHP1)	low_WBC_count	380344	1857	2976	11	14.5	0.76	0.28	0.73	0.37	1.27	1104	0.80
15q11q13dup_BP3_BP5	low_WBC_count	378496	9	2965	0	0.1	0.00	0.30	7.14	0.06	57.03	1149	0.81
3q29_del	low_WBC_count	378496	9	2965	0	0.1	0.00	0.30	7.04	0.05	55.64	1160	0.81
15q13.3del(CHRNA7)	low_WBC_count	378497	10	2965	0	0.1	0.00	0.31	6.71	0.05	52.32	1188	0.81
17q11.2_del_NF1	low_WBC_count	378496	9	2965	0	0.1	0.00	0.32	6.35	0.05	50.20	1231	0.82
1q21.1_dup	low_WBC_count	378653	166	2967	2	1.3	1.53	0.35	1.93	0.40	5.49	1314	0.83
WBS_dup	low_WBC_count	378500	13	2965	0	0.1	0.00	0.37	5.05	0.04	38.12	1368	0.85
22q11.2distal_dup	low_WBC_count	378499	12	2965	0	0.1	0.00	0.38	4.90	0.04	37.36	1394	0.85
15q11q13del_BP3-BP4	low_WBC_count	378501	14	2965	0	0.1	0.00	0.39	4.62	0.04	34.65	1447	0.85
PWS_dup	low_WBC_count	378503	16	2965	0	0.1	0.00	0.41	4.28	0.03	31.73	1495	0.86
17q12_dup	low_WBC_count	378585	98	2966	1	0.8	1.30	0.43	2.06	0.24	7.48	1553	0.87
16p12.1_dup	low_WBC_count	378677	190	2967	2	1.5	1.34	0.45	1.68	0.35	4.76	1622	0.88
TAR_dup	low_WBC_count	378891	404	2969	4	3.2	1.26	0.45	1.46	0.49	3.27	1623	0.88
15q11.2_del	low_WBC_count	380043	1556	2979	14	12.2	1.15	0.48	1.21	0.69	1.95	1714	0.88
15q13.3_dup	low_WBC_count	378709	222	2967	2	1.7	1.15	0.54	1.51	0.32	4.28	1863	0.91
7q11.23_dup_distal	low_WBC_count	378510	23	2965	0	0.2	0.00	0.55	2.72	0.02	19.58	1877	0.91
2q11.2_dup	low_WBC_count	378513	26	2965	0	0.2	0.00	0.59	2.40	0.02	17.16	1989	0.93
16p11.2distal_dup	low_WBC_count	378613	126	2966	1	1.0	1.01	0.60	1.59	0.18	5.72	2012	0.94
2q11.2_del	low_WBC_count	378516	29	2965	0	0.2	0.00	0.61	2.30	0.02	16.38	2025	0.94
16p12.1_del	low_WBC_count	378720	233	2967	2	1.8	1.09	0.64	1.36	0.29	3.85	2130	0.95
17p12_CMT1A_dup	low_WBC_count	378603	116	2965	0	0.9	0.00	0.65	0.56	0.00	3.80	2140	0.95
16p11.2_dup	low_WBC_count	378617	130	2966	1	1.0	0.98	0.65	1.49	0.17	5.37	2152	0.95
15q11.2_dup	low_WBC_count	380388	1901	2981	16	14.9	1.07	0.68	1.11	0.65	1.74	2240	0.95
1q21.1_del	low_WBC_count	378593	106	2965	0	0.8	0.00	0.71	0.61	0.00	4.19	2307	0.96
10q11.21q11.23_dup	low_WBC_count	378527	40	2965	0	0.3	0.00	0.71	1.77	0.01	12.40	2320	0.96
15q13.3_del	low_WBC_count	378524	37	2965	0	0.3	0.00	0.72	1.73	0.01	12.19	2357	0.96
2q21.1_del	low_WBC_count	378527	40	2965	0	0.3	0.00	0.76	1.60	0.01	11.19	2463	0.97
NRXN1_del	low_WBC_count	378643	156	2966	1	1.2	0.82	0.77	1.28	0.15	4.60	2500	0.97
15q11q13dup_BP3_BP4	low_WBC_count	378537	50	2965	0	0.4	0.00	0.84	1.35	0.01	9.41	2710	0.97
16p13.11_dup	low_WBC_count	379264	777	2971	6	6.1	0.99	0.85	1.08	0.45	2.15	2729	0.97
17p12_HNPP_del	low_WBC_count	378705	218	2966	1	1.7	0.59	0.87	0.88	0.10	3.14	2786	0.98
16p11.2distal_del	low_WBC_count	378541	54	2965	0	0.4	0.00	0.88	1.25	0.01	8.67	2813	0.98
13q12.12_dup	low_WBC_count	378698	211	2966	1	1.7	0.61	0.92	0.92	0.11	3.29	2919	0.98
2q13_dup	low_WBC_count	378558	71	2965	0	0.6	0.00	0.96	0.93	0.01	6.43	3037	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
17p12_CMT1A_dup	MI_derived2017	367697	105	13871	11	4.0	2.61	0.0032	2.96	1.48	5.40	65	0.15
22q11.2_dup	MI_derived2017	367840	248	13878	18	9.4	1.86	0.014	1.95	1.16	3.10	147	0.29
16p12.1_del	MI_derived2017	367812	220	13875	15	8.3	1.76	0.025	1.94	1.09	3.19	217	0.36
TAR_del	MI_derived2017	367655	63	13866	6	2.4	2.39	0.030	2.89	1.12	6.31	243	0.38
22q11.2distal_dup	MI_derived2017	367602	10	13862	2	0.4	4.59	0.049	5.49	1.00	20.61	327	0.47
10q11.21q11.23_dup	MI_derived2017	367629	37	13863	3	1.4	2.06	0.090	3.09	0.81	8.49	489	0.58
2q13del(NPHP1)	MI_derived2017	369823	2231	13959	99	84.2	1.17	0.097	1.20	0.97	1.46	515	0.59
1q21.1_dup	MI_derived2017	367750	158	13870	10	6.0	1.64	0.11	1.77	0.87	3.21	542	0.62
PWS_dup	MI_derived2017	367607	15	13861	1	0.6	1.72	0.17	4.42	0.45	20.57	738	0.70
3q29_del	MI_derived2017	367600	8	13861	1	0.3	3.06	0.17	4.42	0.45	22.01	746	0.71
15q11.2_del	MI_derived2017	369112	1520	13910	50	57.3	0.88	0.18	0.83	0.61	1.09	771	0.72
TAR_dup	MI_derived2017	367980	388	13880	20	14.6	1.35	0.18	1.38	0.85	2.12	787	0.73
17q12_del	MI_derived2017	367598	6	13861	1	0.2	3.93	0.20	4.02	0.40	21.75	834	0.75
2q13_del	MI_derived2017	367643	51	13860	0	1.9	0.00	0.20	0.24	0.00	1.72	842	0.74
2q13dup(NPHP1)	MI_derived2017	369401	1809	13919	59	68.2	0.87	0.23	0.85	0.65	1.10	933	0.77
13q12.12_dup	MI_derived2017	367800	208	13864	4	7.8	0.52	0.24	0.60	0.20	1.36	976	0.78
15q11q13dup_BP3_BP4	MI_derived2017	367639	47	13863	3	1.8	1.65	0.26	2.01	0.54	5.37	1036	0.79
2q21.1_del	MI_derived2017	367632	40	13860	0	1.5	0.00	0.26	0.28	0.00	2.00	1042	0.80
15q13.3dup(CHRNA7)	MI_derived2017	370345	2753	13950	90	103.7	0.87	0.27	0.89	0.71	1.09	1053	0.80
13q12.12_del	MI_derived2017	367671	79	13865	5	3.0	1.64	0.28	1.69	0.61	3.77	1100	0.80
NRXN1_del	MI_derived2017	367746	154	13863	3	5.8	0.53	0.31	0.60	0.17	1.52	1202	0.82
15q24_dup	MI_derived2017	367600	8	13860	0	0.3	0.00	0.41	4.31	0.03	35.92	1499	0.86
15q13.3_dup	MI_derived2017	367809	217	13867	7	8.2	0.86	0.43	0.75	0.33	1.47	1539	0.87
3q29_dup	MI_derived2017	367597	5	13860	0	0.19	0.00	0.43	4.09	0.03	37.35	1547	0.87
7q11.23_dup_distal	MI_derived2017	367614	22	13861	1	0.8	1.20	0.48	1.95	0.21	8.16	1712	0.88
22q11.2distal_del	MI_derived2017	367597	5	13860	0	0.2	0.00	0.53	2.95	0.02	29.86	1845	0.90
2q21.1_dup	MI_derived2017	367646	54	13861	1	2.0	0.50	0.55	0.63	0.07	2.39	1896	0.92
16p11.2_del	MI_derived2017	367692	100	13863	3	3.8	0.80	0.58	0.74	0.20	1.93	1959	0.93
1q21.1_del	MI_derived2017	367694	102	13864	4	3.8	1.04	0.63	1.28	0.42	3.00	2087	0.94
Potocki Lupski	MI_derived2017	367597	5	13860	0	0.2	0.00	0.67	2.06	0.01	23.10	2202	0.95
2q13_dup	MI_derived2017	367660	68	13863	3	2.6	1.16	0.68	1.27	0.35	3.33	2229	0.95
8p23.1_dup	MI_derived2017	367597	5	13860	0	0.2	0.00	0.69	1.92	0.01	18.84	2271	0.95
16p13.11_dup	MI_derived2017	368345	753	13890	30	28.4	1.05	0.69	1.08	0.73	1.53	2276	0.95
16p11.2distal_dup	MI_derived2017	367716	124	13863	3	4.7	0.65	0.70	0.81	0.22	2.06	2285	0.96
2q11.2_dup	MI_derived2017	367618	26	13860	0	1.0	0.00	0.72	0.62	0.00	4.57	2328	0.96
15q11q13dup_BP3_BP5	MI_derived2017	367601	9	13860	0	0.3	0.00	0.73	1.73	0.01	15.30	2389	0.96
16p12.1_dup	MI_derived2017	367777	185	13867	7	7.0	1.00	0.76	1.12	0.49	2.21	2470	0.97
15q13.3_del	MI_derived2017	367628	36	13861	1	1.4	0.74	0.76	0.78	0.09	3.05	2473	0.97
13q12dup_CRYL1	MI_derived2017	367602	10	13860	0	0.4	0.00	0.77	1.58	0.01	14.02	2501	0.97
16p11.2distal_del	MI_derived2017	367644	52	13862	2	2.0	1.02	0.80	1.19	0.24	3.57	2579	0.97
2q11.2_del	MI_derived2017	367620	28	13861	1	1.1	0.95	0.81	1.24	0.14	4.96	2610	0.97
10q23_dup	MI_derived2017	367599	7	13860	0	0.3	0.00	0.82	1.45	0.01	14.14	2627	0.97
15q11.2_dup	MI_derived2017	369438	1846	13931	71	69.6	1.02	0.82	1.03	0.80	1.30	2629	0.97
15q11q13del_BP3-BP4	MI_derived2017	367606	14	13860	0	0.5	0.00	0.83	1.39	0.01	10.76	2671	0.97
16p13.11_del	MI_derived2017	367712	120	13864	4	4.5	0.89	0.84	1.11	0.36	2.61	2686	0.97
10q11.21q11.23_del	MI_derived2017	367646	54	13862	2	2.0	0.98	0.85	1.14	0.23	3.49	2732	0.97
22q11.2_del	MI_derived2017	367602	10	13860	0	0.4	0.00	0.94	1.12	0.01	9.92	2978	0.99
WBS_dup	MI_derived2017	367605	13	13860	0	0.5	0.00	0.95	0.92	0.01	7.51	3021	0.99
13q12del_CRYL1	MI_derived2017	367944	352	13871	11	13.3	0.83	0.96	0.98	0.51	1.71	3038	0.99
17p12_HNPP_del	MI_derived2017	367803	211	13868	8	8.0	1.01	0.96	1.02	0.47	1.92	3040	0.99
17q11.2_del_NF1	MI_derived2017	367601	9	13860	0	0.3	0.00	0.96	0.94	0.01	8.00	3049	0.99
16p11.2_dup	MI_derived2017	367718	126	13865	5	4.8	1.05	0.98	1.01	0.37	2.21	3088	1.00
15q13.3del(CHRNA7)	MI_derived2017	367602	10	13860	0	0.4	0.00	0.99	0.98	0.01	8.20	3102	1.00
17q12_dup	MI_derived2017	367687	95	13864	4	3.6	1.11	1.00	1.00	0.33	2.34	3126	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
13q12dup_CRYL1	migraine	367782	8	13680	2	0.3	5.58	0.020	7.98	1.49	29.66	178	0.35
16p11.2_del	migraine	367869	95	13686	8	3.5	2.17	0.020	2.59	1.18	4.98	183	0.35
13q12del_CRYL1	migraine	368131	357	13684	6	13.3	0.46	0.030	0.47	0.19	0.94	242	0.39
3q29_dup	migraine	367778	4	13679	1	0.15	5.58	0.040	11.94	1.16	67.58	289	0.43
15q13.3_dup	migraine	367996	222	13680	2	8.3	0.25	0.048	0.35	0.07	0.99	321	0.47
Potocki_Lupski	migraine	367778	4	13679	1	0.1	5.58	0.063	8.90	0.86	52.19	373	0.53
16p12.1_dup	migraine	367963	189	13681	3	7.0	0.44	0.12	0.47	0.13	1.17	576	0.63
10q11.21q11.23_dup	migraine	367811	37	13681	3	1.4	2.09	0.12	2.71	0.73	7.20	603	0.63
15q24_dup	migraine	367781	7	13679	1	0.3	3.49	0.16	4.38	0.46	20.42	733	0.70
15q11.2_del	migraine	369299	1525	13723	45	56.7	0.80	0.17	0.82	0.60	1.08	739	0.70
15q11q13dup_BP3_BP4	migraine	367821	47	13681	3	1.7	1.67	0.18	2.34	0.64	6.11	761	0.72
13q12.12_dup	migraine	367982	208	13682	4	7.7	0.53	0.18	0.56	0.19	1.26	775	0.72
16p13.11_dup	migraine	368536	762	13699	21	28.3	0.75	0.19	0.76	0.48	1.14	821	0.74
1q21.1_dup	migraine	367933	159	13687	9	5.9	1.49	0.20	1.58	0.76	2.90	843	0.74
16p11.2distal_del	migraine	367828	54	13678	0	2.0	0.00	0.22	0.25	0.00	1.78	901	0.76
15q11q13del_BP3-BP4	migraine	367787	13	13679	1	0.5	1.99	0.26	3.11	0.34	13.01	1022	0.79
17q12_dup	migraine	367872	98	13679	1	3.6	0.28	0.28	0.46	0.05	1.66	1082	0.80
22q11.2_dup	migraine	368027	253	13691	13	9.4	1.36	0.31	1.36	0.73	2.31	1201	0.81
13q12.12_del	migraine	367857	83	13679	1	3.1	0.33	0.32	0.48	0.06	1.77	1233	0.82
15q13.3_del	migraine	367809	35	13680	2	1.3	1.51	0.34	2.01	0.41	6.12	1276	0.83
16p11.2_dup	migraine	367899	125	13684	6	4.6	1.28	0.37	1.46	0.59	2.99	1385	0.85
2q21.1_del	migraine	367812	38	13680	2	1.4	1.39	0.41	1.81	0.37	5.43	1488	0.86
1q21.1_del	migraine	367878	104	13680	2	3.9	0.53	0.42	0.62	0.13	1.78	1519	0.87
16p12.1_del	migraine	368003	229	13684	6	8.5	0.71	0.43	0.74	0.30	1.49	1558	0.87
16p13.11_del	migraine	367892	118	13684	6	4.4	1.35	0.47	1.36	0.55	2.77	1667	0.88
8p23.1_dup	migraine	367779	5	13678	0	0.2	0.00	0.53	2.91	0.02	27.26	1849	0.90
17p12_CMT1A_dup	migraine	367885	111	13683	5	4.1	1.20	0.56	1.30	0.48	2.81	1927	0.91
NRXN1_del	migraine	367927	153	13682	4	5.7	0.71	0.57	0.77	0.26	1.75	1938	0.92
2q13_dup	migraine	367842	68	13681	3	2.5	1.18	0.58	1.38	0.38	3.53	1951	0.93
2q11.2_del	migraine	367803	29	13678	0	1.1	0.00	0.58	0.50	0.00	3.58	1966	0.93
2q11.2_dup	migraine	367800	26	13678	0	1.0	0.00	0.60	0.51	0.00	3.67	2002	0.93
TAR_del	migraine	367840	66	13681	3	2.5	1.21	0.61	1.35	0.37	3.44	2024	0.94
15q11.2_dup	migraine	369618	1844	13751	73	68.6	1.06	0.61	1.06	0.83	1.34	2032	0.94
2q13_del	migraine	367823	49	13680	2	1.8	1.09	0.61	1.42	0.29	4.20	2046	0.94
7q11.23_dup_distal	migraine	367797	23	13678	0	0.9	0.00	0.62	0.53	0.00	3.83	2060	0.94
22q11.2distal_del	migraine	367779	5	13678	0	0.2	0.00	0.64	2.15	0.02	19.91	2132	0.94
17q12_del	migraine	367781	7	13678	0	0.3	0.00	0.65	2.09	0.02	18.51	2157	0.95
10q23_dup	migraine	367781	7	13678	0	0.3	0.00	0.68	1.94	0.01	16.92	2243	0.95
PWS_dup	migraine	367790	16	13678	0	0.6	0.00	0.72	0.62	0.00	4.66	2342	0.96
16p11.2distal_dup	migraine	367896	122	13683	5	4.5	1.10	0.72	1.17	0.44	2.52	2344	0.96
17q11.2_del_NF1	migraine	367783	9	13678	0	0.3	0.00	0.74	1.68	0.01	13.79	2406	0.96
15q13.3del(CHRNA7)	migraine	367784	10	13678	0	0.4	0.00	0.78	1.55	0.01	12.72	2526	0.97
3q29_del	migraine	367783	9	13678	0	0.3	0.00	0.79	1.52	0.01	12.47	2555	0.97
10q11.21q11.23_del	migraine	367828	54	13680	2	2.0	1.00	0.80	1.19	0.24	3.49	2584	0.97
2q21.1_dup	migraine	367828	54	13679	1	2.0	0.51	0.82	0.83	0.09	3.09	2643	0.97
TAR_dup	migraine	368169	395	13691	13	14.7	0.89	0.84	0.95	0.52	1.56	2702	0.97
15q11q13dup_BP3_BP5	migraine	367783	9	13678	0	0.3	0.00	0.87	1.29	0.01	10.50	2780	0.98
2q13dup(NPHP1)	migraine	369577	1803	13743	65	67.0	0.97	0.88	0.98	0.76	1.25	2810	0.98
2q13del(NPHP1)	migraine	370021	2247	13761	83	83.6	0.99	0.88	0.98	0.78	1.22	2816	0.98
22q11.2_del	migraine	367784	10	13678	0	0.4	0.00	0.90	1.22	0.01	9.98	2865	0.98
22q11.2distal_dup	migraine	367786	12	13678	0	0.4	0.00	0.94	1.13	0.01	8.80	2967	0.99
17p12_HNPP_del	migraine	367986	212	13685	7	7.9	0.89	0.94	0.97	0.43	1.88	2996	0.99
15q13.3dup(CHRNA7)	migraine	370516	2742	13779	101	102.0	0.99	0.95	0.99	0.81	1.21	3014	0.99
WBS_dup	migraine	367787	13	13678	0	0.5	0.00	0.99	1.01	0.01	7.87	3116	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
8p23.1_dup	nasal	378264	4	3193	1	0.0	23.90	0.0071	34.71	3.42	194.16	96	0.23
2q13del(NPHP1)	nasal	380560	2300	3222	30	19.5	1.54	0.022	1.57	1.07	2.20	202	0.34
13q12del_CRYL1	nasal	378623	363	3192	0	3.1	0.00	0.072	0.17	0.00	1.12	416	0.54
2q11.2_del	nasal	378288	28	3193	1	0.2	4.12	0.080	6.62	0.74	25.36	452	0.56
10q11.21q11.23_dup	nasal	378299	39	3193	1	0.3	2.99	0.13	4.85	0.55	18.16	628	0.64
22q11.2_dup	nasal	378522	262	3196	4	2.2	1.80	0.15	2.17	0.72	4.89	686	0.68
16p12.1_dup	nasal	378449	189	3195	3	1.6	1.87	0.18	2.27	0.63	5.64	778	0.72
2q13_del	nasal	378310	50	3193	1	0.4	2.34	0.19	3.68	0.42	13.62	815	0.74
22q11.2distal_del	nasal	378265	5	3192	0	0.0	0.00	0.20	14.10	0.11	127.76	825	0.74
16p13.11_del	nasal	378382	122	3194	2	1.0	1.93	0.21	2.53	0.53	7.23	867	0.75
16p11.2distal_dup	nasal	378385	125	3194	2	1.1	1.88	0.21	2.52	0.53	7.21	868	0.75
10q11.21q11.23_del	nasal	378315	55	3193	1	0.5	2.13	0.21	3.45	0.39	12.74	880	0.75
Potocki_Lupski	nasal	378265	5	3192	0	0.0	0.00	0.22	11.89	0.09	106.96	893	0.76
15q13.3_dup	nasal	378484	224	3192	0	1.9	0.00	0.22	0.26	0.00	1.75	894	0.76
3q29_dup	nasal	378265	5	3192	0	0.04	0.00	0.23	10.64	0.08	97.31	942	0.78
15q24_dup	nasal	378268	8	3192	0	0.1	0.00	0.24	9.84	0.08	79.73	975	0.78
17q12_del	nasal	378267	7	3192	0	0.1	0.00	0.28	7.90	0.06	66.90	1096	0.80
17p12_HNPP_del	nasal	378476	216	3195	3	1.8	1.64	0.28	1.89	0.53	4.69	1105	0.80
10q23_dup	nasal	378267	7	3192	0	0.1	0.00	0.28	7.74	0.06	64.38	1110	0.80
15q11q13dup_BP3_BP5	nasal	378269	9	3192	0	0.1	0.00	0.30	7.15	0.06	57.15	1147	0.81
3q29_del	nasal	378269	9	3192	0	0.1	0.00	0.30	7.13	0.06	56.79	1151	0.81
22q11.2_del	nasal	378270	10	3192	0	0.1	0.00	0.31	6.69	0.05	52.94	1192	0.81
13q12dup_CRYL1	nasal	378270	10	3192	0	0.1	0.00	0.32	6.47	0.05	50.89	1216	0.82
15q13.3del(CHRNA7)	nasal	378270	10	3192	0	0.1	0.00	0.33	6.17	0.05	48.35	1245	0.82
17q11.2_del_NF1	nasal	378269	9	3192	0	0.1	0.00	0.36	5.21	0.04	42.14	1351	0.85
2q13dup(NPHP1)	nasal	380116	1856	3204	12	15.6	0.77	0.38	0.79	0.43	1.31	1409	0.85
WBS_dup	nasal	378273	13	3192	0	0.1	0.00	0.39	4.74	0.04	36.25	1422	0.85
1q21.1_dup	nasal	378428	168	3192	0	1.4	0.00	0.39	0.36	0.00	2.49	1456	0.85
22q11.2distal_dup	nasal	378272	12	3192	0	0.1	0.00	0.40	4.56	0.04	35.02	1461	0.85
PWS_dup	nasal	378276	16	3192	0	0.1	0.00	0.40	4.47	0.03	33.70	1472	0.85
15q11q13del_BP3-BP4	nasal	378274	14	3192	0	0.1	0.00	0.40	4.41	0.03	33.23	1479	0.85
16p11.2_del	nasal	378362	102	3193	1	0.9	1.16	0.44	2.05	0.23	7.42	1566	0.87
17q12_dup	nasal	378358	98	3193	1	0.8	1.21	0.54	1.73	0.20	6.28	1861	0.91
16p11.2_dup	nasal	378391	131	3192	0	1.1	0.00	0.54	0.47	0.00	3.21	1868	0.91
7q11.23_dup_distal	nasal	378283	23	3192	0	0.2	0.00	0.56	2.60	0.02	18.73	1928	0.91
17p12_CMT1A_dup	nasal	378375	115	3193	1	1.0	1.03	0.59	1.60	0.18	5.80	1996	0.93
2q11.2_dup	nasal	378286	26	3192	0	0.2	0.00	0.61	2.28	0.02	16.41	2040	0.94
1q21.1_del	nasal	378366	106	3192	0	0.9	0.00	0.67	0.58	0.00	3.95	2204	0.95
15q13.3_del	nasal	378297	37	3192	0	0.3	0.00	0.73	1.70	0.01	12.02	2385	0.96
15q11.2_dup	nasal	380163	1903	3206	14	16.0	0.87	0.75	0.92	0.52	1.49	2438	0.97
15q11.2_del	nasal	379816	1556	3206	14	13.1	1.07	0.75	1.09	0.62	1.76	2441	0.97
13q12.12_del	nasal	378344	84	3192	0	0.7	0.00	0.77	0.68	0.01	4.69	2499	0.97
16p12.1_del	nasal	378494	234	3193	1	2.0	0.51	0.78	0.80	0.09	2.87	2527	0.97
2q21.1_del	nasal	378300	40	3192	0	0.3	0.00	0.83	1.37	0.01	9.67	2679	0.97
13q12.12_dup	nasal	378471	211	3193	1	1.8	0.56	0.84	0.85	0.10	3.05	2695	0.97
15q13.3dup(CHRNA7)	nasal	381081	2821	3214	22	23.8	0.92	0.86	0.96	0.62	1.42	2770	0.98
2q13_dup	nasal	378331	71	3192	0	0.6	0.00	0.88	0.81	0.01	5.60	2811	0.98
NRXN1_del	nasal	378416	156	3193	1	1.3	0.76	0.88	1.13	0.13	4.07	2818	0.98
15q11q13dup_BP3_BP4	nasal	378310	50	3192	0	0.4	0.00	0.91	1.19	0.01	8.27	2889	0.98
TAR_del	nasal	378329	69	3192	0	0.6	0.00	0.92	0.88	0.01	6.07	2941	0.98
TAR_dup	nasal	378665	405	3195	3	3.4	0.88	0.94	1.04	0.29	2.56	2995	0.99
16p11.2distal_del	nasal	378314	54	3192	0	0.5	0.00	0.96	1.07	0.01	7.48	3039	0.99
2q21.1_dup	nasal	378315	55	3192	0	0.5	0.00	0.97	1.05	0.01	7.33	3070	0.99
16p13.11_dup	nasal	379037	777	3198	6	6.6	0.92	0.98	1.01	0.42	2.01	3086	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
17p12_CMT1A_dup	neuropathies	365609	60	15959	56	2.6	11.58	3.94E-124	21.75	15.02	31.46	1	1.23E-120
17p12_HNPP_del	neuropathies	365743	194	15928	25	8.4	2.74	5.44E-06	3.02	1.95	4.49	10	0.0017
3q29_dup	neuropathies	365552	3	15905	2	0.13	9.59	0.0019	24.65	3.94	131.53	54	0.11
15q11.2_dup	neuropathies	367359	1810	16010	107	78.9	1.34	0.0036	1.36	1.11	1.64	70	0.16
10q11.21q11.23_dup	neuropathies	365585	36	15907	4	1.6	2.40	0.043	3.25	1.04	7.88	304	0.45
15q13.3_del	neuropathies	365582	33	15907	4	1.4	2.59	0.067	2.87	0.92	6.99	387	0.54
PWS_dup	neuropathies	365563	14	15905	2	0.6	3.00	0.076	4.23	0.83	13.94	433	0.55
10q23_dup	neuropathies	365555	6	15904	1	0.3	3.43	0.11	5.83	0.60	28.55	554	0.62
22q11.2_dup	neuropathies	365799	250	15919	16	10.9	1.44	0.13	1.50	0.88	2.40	647	0.64
1q21.1_dup	neuropathies	365706	157	15914	11	6.8	1.57	0.14	1.62	0.84	2.83	670	0.66
TAR_dup	neuropathies	365935	386	15925	22	16.8	1.29	0.17	1.37	0.87	2.05	742	0.71
15q13.3del(CHRNA7)	neuropathies	365558	9	15904	1	0.4	2.40	0.19	3.86	0.41	16.96	814	0.74
1q21.1_del	neuropathies	365648	99	15910	7	4.3	1.58	0.20	1.70	0.74	3.34	823	0.74
15q11q13dup_BP3_BP5	neuropathies	365557	8	15904	1	0.3	2.67	0.20	3.85	0.41	17.37	824	0.74
16p12.1_dup	neuropathies	365730	181	15914	11	7.9	1.37	0.27	1.43	0.74	2.48	1052	0.80
2q11.2_del	neuropathies	365576	27	15905	2	1.2	1.65	0.28	2.22	0.45	6.78	1094	0.80
10q11.21q11.23_del	neuropathies	365601	52	15907	4	2.3	1.71	0.28	1.78	0.58	4.20	1095	0.80
15q11q13del_BP3-BP4	neuropathies	365562	13	15904	1	0.6	1.71	0.31	2.71	0.29	11.15	1194	0.81
WBS_dup	neuropathies	365561	12	15904	1	0.5	1.85	0.34	2.56	0.28	10.83	1281	0.82
NRXN1_del	neuropathies	365702	153	15907	4	6.7	0.61	0.39	0.68	0.23	1.54	1433	0.85
16p11.2_dup	neuropathies	365673	124	15910	7	5.4	1.28	0.41	1.39	0.60	2.71	1486	0.86
2q13dup(NPHP1)	neuropathies	367346	1797	15974	71	78.1	0.91	0.45	0.91	0.71	1.15	1611	0.87
15q13.3dup(CHRNA7)	neuropathies	368268	2719	16027	124	118.3	1.05	0.50	1.06	0.88	1.27	1763	0.89
13q12del_CRYL1	neuropathies	365899	350	15916	13	15.2	0.86	0.52	0.84	0.45	1.41	1824	0.90
2q11.2_dup	neuropathies	365575	26	15903	0	1.1	0.00	0.53	0.45	0.00	3.25	1835	0.90
13q12.12_del	neuropathies	365631	82	15905	2	3.6	0.57	0.55	0.69	0.14	2.00	1875	0.91
16p11.2distal_del	neuropathies	365602	53	15904	1	2.3	0.44	0.58	0.65	0.07	2.41	1963	0.93
16p11.2distal_dup	neuropathies	365670	121	15909	6	5.3	1.13	0.63	1.23	0.50	2.49	2082	0.94
16p13.11_dup	neuropathies	366302	753	15933	30	32.8	0.92	0.64	0.92	0.63	1.30	2129	0.95
7q11.23_dup_distal	neuropathies	365571	22	15904	1	1.0	1.04	0.65	1.50	0.17	5.84	2155	0.95
8p23.1_dup	neuropathies	365554	5	15903	0	0.2	0.00	0.65	2.08	0.02	18.70	2161	0.95
15q13.3_dup	neuropathies	365763	214	15913	10	9.3	1.07	0.66	1.15	0.58	2.04	2182	0.95
16p11.2_del	neuropathies	365649	100	15906	3	4.4	0.70	0.67	0.80	0.22	2.00	2205	0.95
Potocki_Lupski	neuropathies	365554	5	15903	0	0.2	0.00	0.68	1.96	0.01	17.64	2225	0.95
16p13.11_del	neuropathies	365669	120	15907	4	5.2	0.77	0.69	0.83	0.27	1.89	2260	0.95
15q11q13dup_BP3_BP4	neuropathies	365597	48	15905	2	2.1	0.96	0.70	1.30	0.27	3.83	2290	0.95
2q13_dup	neuropathies	365617	68	15906	3	3.0	1.01	0.72	1.22	0.34	3.11	2369	0.96
22q11.2distal_del	neuropathies	365554	5	15903	0	0.2	0.00	0.75	1.66	0.01	15.47	2437	0.97
17q12_del	neuropathies	365556	7	15903	0	0.3	0.00	0.76	1.63	0.01	13.43	2455	0.96
2q13_del	neuropathies	365598	49	15905	2	2.1	0.94	0.79	1.20	0.25	3.52	2552	0.97
17q12_dup	neuropathies	365645	96	15906	3	4.2	0.73	0.82	0.89	0.25	2.23	2653	0.97
2q21.1_del	neuropathies	365588	39	15904	1	1.7	0.60	0.83	0.84	0.09	3.14	2672	0.97
TAR_del	neuropathies	365616	67	15905	2	2.9	0.70	0.84	0.88	0.18	2.54	2685	0.97
13q12.12_dup	neuropathies	365752	203	15912	9	8.8	1.02	0.84	1.07	0.52	1.94	2691	0.97
22q11.2distal_dup	neuropathies	365561	12	15903	0	0.5	0.00	0.84	0.76	0.01	5.85	2711	0.97
15q24_dup	neuropathies	365557	8	15903	0	0.3	0.00	0.84	1.36	0.01	11.06	2713	0.97
2q13del(NPHP1)	neuropathies	367783	2234	15999	96	97.2	0.99	0.88	0.98	0.80	1.20	2803	0.98
2q21.1_dup	neuropathies	365602	53	15905	2	2.3	0.87	0.88	1.11	0.23	3.25	2804	0.98
17q11.2_del_NF1	neuropathies	365558	9	15903	0	0.4	0.00	0.89	1.23	0.01	9.74	2845	0.98
16p12.1_del	neuropathies	365774	225	15913	10	9.8	1.02	0.90	1.04	0.52	1.83	2885	0.98
3q29_del	neuropathies	365558	9	15903	0	0.4	0.00	0.92	1.17	0.01	9.34	2921	0.98
22q11.2_del	neuropathies	365559	10	15903	0	0.4	0.00	0.94	1.12	0.01	8.81	2968	0.99
13q12dup_CRYL1	neuropathies	365559	10	15903	0	0.4	0.00	0.96	1.07	0.01	8.33	3050	0.99
15q11.2_del	neuropathies	367054	1505	15968	65	65.5	0.99	0.99	1.00	0.77	1.27	3101	1.00



CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2_del	obesity	371699	84	9856	19	2.2	7.15	7.39E-10	6.81	4.01	11.01	3	7.71E-07*
16p12.1_del	obesity	371827	212	9860	23	5.6	3.80	1.11E-07	3.99	2.53	5.99	7	4.95E-05*
16p11.2distal_del	obesity	371660	45	9846	9	1.2	6.46	1.14E-05	7.06	3.27	13.73	13	0.0028*
TAR_dup	obesity	372000	385	9860	23	10.2	2.19	0.00054	2.29	1.47	3.40	36	0.047*
16p13.11_del	obesity	371729	114	9847	10	3.0	3.13	0.0013	3.41	1.70	6.15	42	0.096*
22q11.2distal_del	obesity	371618	3	9839	2	0.1	15.51	0.0038	18.77	2.98	100.40	72	0.17
1q21.1_dup	obesity	371772	157	9848	11	4.2	2.54	0.0086	2.50	1.29	4.36	108	0.25
3q29_dup	obesity	371619	4	9838	1	0.11	7.76	0.021	17.37	1.72	95.51	191	0.35
2q11.2_del	obesity	371641	26	9840	3	0.7	4.01	0.029	4.48	1.20	12.20	241	0.38
8p23.1_dup	obesity	371619	4	9838	1	0.1	7.76	0.033	12.96	1.29	70.70	256	0.41
10q11.21q11.23_dup	obesity	371652	37	9840	3	1.0	2.91	0.046	3.79	1.03	10.01	316	0.46
15q24_dup	obesity	371622	7	9838	1	0.2	4.85	0.070	7.67	0.80	35.48	405	0.54
2q13del(NPHP1)	obesity	373870	2255	9912	75	59.8	1.25	0.075	1.24	0.98	1.56	428	0.55
15q13.3_dup	obesity	371828	213	9848	11	5.6	1.90	0.081	1.78	0.93	3.09	453	0.56
15q13.3_del	obesity	371649	34	9840	3	0.9	3.14	0.11	2.83	0.76	7.52	543	0.62
22q11.2_dup	obesity	371869	254	9849	12	6.7	1.75	0.12	1.63	0.87	2.76	590	0.63
13q12.12_dup	obesity	371825	210	9839	2	5.6	0.37	0.14	0.44	0.09	1.24	659	0.65
NRXN1_del	obesity	371765	150	9844	7	4.0	1.73	0.14	1.84	0.80	3.58	664	0.65
15q13.3del(CHRNA7)	obesity	371624	9	9838	1	0.2	3.88	0.14	4.77	0.51	21.20	673	0.66
22q11.2_del	obesity	371624	9	9838	1	0.2	3.88	0.16	4.49	0.48	19.93	708	0.69
15q11q13del_BP3-BP4	obesity	371628	13	9838	1	0.3	2.77	0.17	4.17	0.45	17.34	743	0.71
16p11.2_dup	obesity	371745	130	9838	1	3.4	0.30	0.20	0.40	0.05	1.46	826	0.74
16p11.2distal_dup	obesity	371741	126	9838	1	3.3	0.31	0.21	0.41	0.05	1.49	873	0.75
2q13_dup	obesity	371686	71	9837	0	1.9	0.00	0.23	0.26	0.00	1.81	926	0.77
2q13dup(NPHP1)	obesity	373443	1828	9877	40	48.3	0.83	0.23	0.83	0.60	1.12	935	0.78
TAR_del	obesity	371681	66	9840	3	1.7	1.69	0.23	2.07	0.57	5.28	940	0.78
15q11.2_dup	obesity	373474	1859	9895	58	49.3	1.17	0.29	1.16	0.88	1.49	1124	0.81
16p13.11_dup	obesity	372373	758	9862	25	20.1	1.24	0.35	1.22	0.80	1.77	1311	0.83
2q11.2_dup	obesity	371640	25	9838	1	0.7	1.49	0.37	2.34	0.26	9.04	1371	0.85
15q13.3dup(CHRNA7)	obesity	374379	2764	9916	79	73.2	1.08	0.38	1.11	0.88	1.38	1410	0.85
15q11q13dup_BP3_BP4	obesity	371665	50	9837	0	1.3	0.00	0.40	0.37	0.00	2.57	1480	0.85
16p12.1_dup	obesity	371804	189	9840	3	5.0	0.61	0.44	0.68	0.19	1.68	1577	0.87
17p12_HNPP_del	obesity	371827	212	9844	7	5.6	1.24	0.47	1.33	0.58	2.56	1660	0.88
Potocki_Lupski	obesity	371620	5	9837	0	0.1	0.00	0.51	3.08	0.02	27.58	1797	0.89
13q12del_CRYL1	obesity	371971	356	9844	7	9.4	0.75	0.60	0.83	0.36	1.59	2003	0.93
17q12_dup	obesity	371711	96	9840	3	2.5	1.18	0.61	1.34	0.37	3.36	2043	0.94
10q23_dup	obesity	371622	7	9837	0	0.2	0.00	0.62	2.26	0.02	19.27	2065	0.94
17q12_del	obesity	371622	7	9837	0	0.2	0.00	0.62	2.24	0.02	18.62	2070	0.94
15q11.2_del	obesity	373147	1532	9875	38	40.5	0.94	0.63	0.93	0.66	1.26	2106	0.94
17q11.2_del_NF1	obesity	371624	9	9837	0	0.2	0.00	0.64	2.12	0.02	16.76	2125	0.95
3q29_del	obesity	371624	9	9837	0	0.2	0.00	0.72	1.75	0.01	13.90	2363	0.96
13q12dup_CRYL1	obesity	371625	10	9837	0	0.3	0.00	0.73	1.70	0.01	13.32	2390	0.96
2q21.1_del	obesity	371654	39	9838	1	1.0	0.97	0.76	1.30	0.15	4.87	2480	0.97
1q21.1_del	obesity	371718	103	9840	3	2.7	1.10	0.77	1.18	0.33	2.97	2488	0.97
15q11q13dup_BP3_BP5	obesity	371624	9	9837	0	0.2	0.00	0.77	1.57	0.01	12.61	2492	0.97
10q11.21q11.23_del	obesity	371670	55	9838	1	1.5	0.69	0.84	0.85	0.10	3.13	2698	0.97
13q12.12_del	obesity	371697	82	9839	2	2.2	0.92	0.86	1.13	0.23	3.25	2753	0.97
7q11.23_dup_distal	obesity	371638	23	9837	0	0.6	0.00	0.91	0.85	0.01	6.13	2887	0.98
17p12_CMT1A_dup	obesity	371728	113	9840	3	3.0	1.00	0.92	1.06	0.29	2.65	2924	0.98
PWS_dup	obesity	371631	16	9837	0	0.4	0.00	0.95	1.10	0.01	8.20	3006	0.99
WBS_dup	obesity	371628	13	9837	0	0.3	0.00	0.95	1.09	0.01	8.39	3023	0.99
22q11.2distal_dup	obesity	371627	12	9837	0	0.3	0.00	0.96	1.07	0.01	8.51	3046	0.99
2q13_del	obesity	371665	50	9838	1	1.3	0.76	0.99	1.01	0.11	3.76	3100	1.00
2q21.1_dup	obesity	371669	54	9838	1	1.4	0.71	1.00	1.00	0.11	3.68	3123	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
15q13.3_del	ocular	359823	31	21666	6	1.9	2.86	0.012	3.52	1.37	7.75	136	0.27
17q12_del	ocular	359797	5	21662	2	0.3	5.03	0.023	7.90	1.39	33.67	207	0.35
2q13_dup	ocular	359863	71	21660	0	4.3	0.00	0.026	0.12	0.00	0.83	222	0.36
1q21.1_del	ocular	359887	95	21671	11	5.7	1.83	0.028	2.14	1.09	3.80	235	0.37
TAR_del	ocular	359861	69	21660	0	4.2	0.00	0.028	0.12	0.00	0.85	236	0.37
3q29_del	ocular	359799	7	21662	2	0.4	3.91	0.042	5.84	1.08	22.24	298	0.44
22q11.2distal_del	ocular	359796	4	21661	1	0.2	3.52	0.086	7.14	0.70	40.03	476	0.57
16p12.1_dup	ocular	359968	176	21676	16	10.6	1.47	0.091	1.60	0.92	2.57	492	0.58
PWS_dup	ocular	359806	14	21662	2	0.8	2.20	0.093	3.92	0.76	13.22	500	0.58
22q11.2_dup	ocular	360038	246	21680	20	14.8	1.32	0.11	1.48	0.91	2.28	541	0.62
10q11.21q11.23_dup	ocular	359828	36	21664	4	2.2	1.76	0.11	2.48	0.79	6.02	545	0.62
15q11q13dup_BP3_BP4	ocular	359837	45	21665	5	2.7	1.76	0.12	2.19	0.80	4.93	592	0.63
2q11.2_dup	ocular	359815	23	21663	3	1.4	2.03	0.13	2.67	0.71	7.34	641	0.64
16p11.2_del	ocular	359887	95	21668	8	5.7	1.37	0.16	1.73	0.79	3.31	718	0.69
2q11.2_del	ocular	359818	26	21663	3	1.6	1.82	0.16	2.46	0.66	6.67	724	0.70
13q12.12_dup	ocular	359988	196	21676	16	11.8	1.33	0.18	1.44	0.83	2.31	779	0.72
22q11.2_del	ocular	359801	9	21661	1	0.5	1.76	0.22	3.50	0.37	15.61	911	0.77
15q13.3_dup	ocular	360008	216	21668	8	13.0	0.63	0.23	0.67	0.31	1.26	932	0.77
NRXN1_del	ocular	359944	152	21665	5	9.1	0.56	0.24	0.62	0.23	1.32	950	0.78
10q11.21q11.23_del	ocular	359843	51	21665	5	3.1	1.57	0.25	1.74	0.64	3.87	1011	0.79
15q13.3del(CHRNA7)	ocular	359801	9	21661	1	0.5	1.76	0.27	3.05	0.32	13.51	1059	0.80
13q12del_CRYL1	ocular	360131	339	21684	24	20.4	1.16	0.27	1.27	0.82	1.88	1067	0.80
15q11.2_del	ocular	361263	1471	21759	99	88.6	1.11	0.29	1.12	0.91	1.36	1134	0.81
13q12dup_CRYL1	ocular	359801	9	21661	1	0.5	1.76	0.30	2.82	0.30	12.43	1162	0.81
TAR_dup	ocular	360173	381	21687	27	22.9	1.17	0.31	1.23	0.82	1.78	1187	0.81
2q13del(NPHP1)	ocular	361979	2187	21803	143	131.7	1.08	0.34	1.09	0.91	1.28	1299	0.83
16p11.2distal_dup	ocular	359915	123	21664	4	7.4	0.55	0.35	0.66	0.22	1.51	1327	0.84
17q11.2_del_NF1	ocular	359800	8	21661	1	0.5	1.96	0.36	2.45	0.26	11.01	1353	0.84
17p12_CMT1A_dup	ocular	359904	112	21664	4	6.7	0.61	0.38	0.67	0.22	1.54	1400	0.85
15q13.3dup(CHRNA7)	ocular	362486	2694	21809	149	162.1	0.92	0.41	0.93	0.79	1.10	1501	0.86
15q11q13del_BP3-BP4	ocular	359805	13	21661	1	0.8	1.26	0.42	2.15	0.23	8.89	1525	0.87
17q12_dup	ocular	359884	92	21667	7	5.5	1.25	0.43	1.37	0.59	2.71	1552	0.87
22q11.2distal_dup	ocular	359803	11	21661	1	0.7	1.47	0.49	1.92	0.21	8.09	1732	0.89
1q21.1_dup	ocular	359953	161	21667	7	9.7	0.73	0.50	0.78	0.34	1.52	1766	0.89
2q21.1_del	ocular	359831	39	21661	1	2.3	0.44	0.52	0.61	0.07	2.29	1829	0.90
16p13.11_del	ocular	359908	116	21668	8	7.0	1.14	0.57	1.23	0.57	2.34	1941	0.92
3q29_dup	ocular	359797	5	21660	0	0.30	0.00	0.65	2.11	0.02	19.42	2153	0.94
2q21.1_dup	ocular	359845	53	21662	2	3.2	0.64	0.70	0.79	0.16	2.31	2298	0.96
16p11.2distal_del	ocular	359843	51	21663	3	3.1	0.98	0.72	1.23	0.34	3.16	2339	0.96
16p13.11_dup	ocular	360534	742	21701	41	44.7	0.92	0.73	0.95	0.68	1.28	2373	0.96
16p12.1_del	ocular	360013	221	21674	14	13.3	1.05	0.74	1.10	0.62	1.80	2396	0.96
WBS_dup	ocular	359805	13	21660	0	0.8	0.00	0.75	0.65	0.01	4.97	2422	0.97
Potocki_Lupski	ocular	359797	5	21660	0	0.3	0.00	0.80	1.49	0.01	13.41	2583	0.97
2q13dup(NPHP1)	ocular	361552	1760	21768	108	106.0	1.02	0.82	1.02	0.84	1.24	2654	0.97
13q12.12_del	ocular	359871	79	21665	5	4.8	1.05	0.83	1.10	0.41	2.40	2661	0.97
17p12_HNPP_del	ocular	359998	206	21673	13	12.4	1.05	0.85	1.05	0.58	1.76	2738	0.97
8p23.1_dup	ocular	359797	5	21660	0	0.3	0.00	0.86	1.32	0.01	11.77	2761	0.97
15q24_dup	ocular	359800	8	21660	0	0.5	0.00	0.86	1.31	0.01	10.84	2762	0.97
7q11.23_dup_distal	ocular	359814	22	21661	1	1.3	0.77	0.89	1.13	0.12	4.39	2847	0.98
10q23_dup	ocular	359799	7	21660	0	0.4	0.00	0.89	1.23	0.01	10.53	2850	0.98
2q13_del	ocular	359841	49	21662	2	2.9	0.69	0.93	0.94	0.19	2.78	2944	0.99
15q11.2_dup	ocular	361602	1810	21767	107	109.0	0.98	0.98	1.00	0.82	1.21	3091	1.00
15q11q13dup_BP3_BP5	ocular	359801	9	21660	0	0.5	0.00	0.99	1.02	0.01	8.26	3109	1.00
16p11.2_dup	ocular	359916	124	21667	7	7.5	0.94	1.00	1.00	0.44	1.96	3125	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2_del	osteoarthritis	312820	73	68735	30	16.0	1.62	0.00031	2.36	1.50	3.63	28	0.035*
17p12_CMT1A_dup	osteoarthritis	312830	83	68738	33	18.2	1.58	0.0029	1.94	1.26	2.93	64	0.14
16p12.1_dup	osteoarthritis	312893	146	68751	46	32.1	1.33	0.025	1.50	1.06	2.10	213	0.36
1q21.1_del	osteoarthritis	312826	79	68732	27	17.4	1.41	0.026	1.70	1.07	2.64	223	0.36
15q11.2_del	osteoarthritis	314063	1316	68959	254	289.0	0.90	0.058	0.88	0.76	1.00	358	0.50
16p12.1_del	osteoarthritis	312929	182	68758	53	40.0	1.25	0.066	1.35	0.98	1.84	382	0.55
2q11.2_del	osteoarthritis	312768	21	68713	8	4.6	1.53	0.078	2.19	0.91	4.82	441	0.56
13q12.12_dup	osteoarthritis	312930	183	68734	29	40.2	0.76	0.11	0.73	0.48	1.07	563	0.63
22q11.2distal_del	osteoarthritis	312750	3	68707	2	0.7	2.22	0.13	4.20	0.63	23.62	630	0.64
15q11q13dup_BP3_BP5	osteoarthritis	312753	6	68708	3	1.3	1.85	0.15	2.95	0.65	11.50	682	0.68
17p12_HNPP_del	osteoarthritis	312933	186	68738	33	40.9	0.84	0.15	0.76	0.52	1.10	694	0.68
15q13.3_del	osteoarthritis	312774	27	68715	10	5.9	1.50	0.15	1.74	0.80	3.53	699	0.69
15q13.3del(CHRNA7)	osteoarthritis	312754	7	68708	3	1.5	1.67	0.16	2.79	0.65	9.86	705	0.69
2q13dup(NPHP1)	osteoarthritis	314258	1511	69062	357	332.1	1.06	0.16	1.09	0.97	1.23	725	0.70
22q11.2_dup	osteoarthritis	312959	212	68759	54	46.6	1.13	0.16	1.25	0.91	1.69	734	0.70
15q11q13dup_BP3_BP4	osteoarthritis	312786	39	68716	11	8.6	1.22	0.22	1.56	0.75	3.01	908	0.77
10q23_dup	osteoarthritis	312752	5	68707	2	1.1	1.59	0.23	2.84	0.48	13.22	919	0.77
3q29_del	osteoarthritis	312756	9	68705	0	2.0	0.00	0.23	0.24	0.00	1.96	921	0.77
2q13del(NPHP1)	osteoarthritis	314631	1884	69151	446	414.1	1.06	0.23	1.07	0.96	1.19	929	0.77
2q11.2_dup	osteoarthritis	312771	24	68707	2	5.3	0.43	0.24	0.49	0.10	1.54	981	0.78
2q21.1_del	osteoarthritis	312777	30	68715	10	6.6	1.39	0.25	1.56	0.72	3.12	1000	0.78
15q13.3_dup	osteoarthritis	312939	192	68737	32	42.2	0.79	0.26	0.81	0.54	1.17	1040	0.80
1q21.1_dup	osteoarthritis	312890	143	68730	25	31.4	0.83	0.30	0.80	0.51	1.21	1172	0.81
13q12.12_del	osteoarthritis	312819	72	68717	12	15.8	0.79	0.31	0.74	0.38	1.31	1195	0.81
16p11.2distal_dup	osteoarthritis	312848	101	68731	26	22.2	1.14	0.31	1.27	0.79	1.95	1204	0.81
22q11.2distal_dup	osteoarthritis	312758	11	68706	1	2.4	0.46	0.33	0.46	0.05	1.97	1246	0.82
17q12_del	osteoarthritis	312754	7	68705	0	1.5	0.00	0.38	0.33	0.00	2.85	1408	0.85
2q21.1_dup	osteoarthritis	312795	48	68712	7	10.5	0.71	0.43	0.74	0.31	1.52	1544	0.87
TAR_dup	osteoarthritis	313091	344	68769	64	75.6	0.87	0.43	0.90	0.68	1.17	1550	0.87
TAR_del	osteoarthritis	312807	60	68714	9	13.2	0.72	0.43	0.76	0.36	1.46	1554	0.87
WBS_dup	osteoarthritis	312759	12	68706	1	2.6	0.43	0.47	0.55	0.06	2.42	1678	0.88
Potocki_Lupski	osteoarthritis	312752	5	68705	0	1.1	0.00	0.47	0.39	0.00	3.52	1679	0.88
16p13.11_del	osteoarthritis	312846	99	68730	25	21.7	1.12	0.48	1.18	0.74	1.82	1686	0.88
17q12_dup	osteoarthritis	312827	80	68724	19	17.6	1.07	0.50	1.20	0.70	1.95	1753	0.89
15q13.3dup(CHRNA7)	osteoarthritis	315078	2331	69217	512	512.1	1.00	0.54	1.03	0.93	1.14	1852	0.91
10q11.21q11.23_dup	osteoarthritis	312783	36	68709	4	7.9	0.56	0.54	0.74	0.23	1.84	1870	0.91
16p11.2distal_del	osteoarthritis	312794	47	68712	7	10.3	0.72	0.59	0.81	0.34	1.67	1984	0.93
PWS_dup	osteoarthritis	312762	15	68706	1	3.3	0.35	0.60	0.65	0.07	2.76	2014	0.94
10q11.21q11.23_del	osteoarthritis	312791	44	68717	12	9.7	1.19	0.60	1.19	0.60	2.22	2016	0.94
NRXN1_del	osteoarthritis	312875	128	68734	29	28.1	1.03	0.63	1.11	0.72	1.65	2089	0.94
17q11.2_del_NF1	osteoarthritis	312755	8	68706	1	1.8	0.62	0.63	0.66	0.07	3.01	2103	0.94
13q12dup_CRYL1	osteoarthritis	312756	9	68706	1	2.0	0.56	0.67	0.70	0.07	3.12	2221	0.95
8p23.1_dup	osteoarthritis	312751	4	68706	1	0.9	1.11	0.73	1.40	0.14	7.78	2382	0.96
13q12del_CRYL1	osteoarthritis	313048	301	68767	62	66.1	0.95	0.79	1.04	0.78	1.36	2545	0.97
2q13_del	osteoarthritis	312791	44	68712	7	9.7	0.76	0.79	0.90	0.37	1.88	2550	0.97
16p11.2_dup	osteoarthritis	312856	109	68727	22	23.9	0.93	0.80	0.94	0.58	1.48	2590	0.97
3q29_dup	osteoarthritis	312752	5	68705	0	1.10	0.00	0.83	0.73	0.01	7.23	2675	0.97
15q11q13del_BP3-BP4	osteoarthritis	312759	12	68707	2	2.6	0.79	0.83	1.16	0.22	4.01	2681	0.97
7q11.23_dup_distal	osteoarthritis	312766	19	68709	4	4.2	0.97	0.85	1.11	0.34	2.91	2737	0.97
15q24_dup	osteoarthritis	312754	7	68706	1	1.5	0.69	0.88	1.16	0.12	5.78	2809	0.98
15q11.2_dup	osteoarthritis	314323	1576	69046	341	346.2	0.99	0.90	0.99	0.88	1.12	2871	0.98
16p13.11_dup	osteoarthritis	313390	643	68845	140	141.3	0.99	0.90	1.01	0.84	1.22	2877	0.98
2q13_dup	osteoarthritis	312806	59	68717	12	13.0	0.94	0.92	1.03	0.53	1.87	2917	0.98
22q11.2_del	osteoarthritis	312756	9	68706	1	2.0	0.56	0.97	1.03	0.11	4.73	3066	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q13del(NPHP1)	osteoporosis	372095	2236	11687	94	70.2	1.33	0.0073	1.35	1.09	1.66	100	0.23
10q11.21q11.23_dup	osteoporosis	369896	37	11596	3	1.2	2.47	0.027	4.77	1.23	13.68	231	0.37
22q11.2distal_del	osteoporosis	369863	4	11594	1	0.1	6.58	0.031	16.66	1.37	118.11	247	0.39
Potocki_Lupski	osteoporosis	369863	4	11594	1	0.1	6.58	0.043	11.56	1.10	70.25	303	0.45
16p13.11_del	osteoporosis	369975	116	11601	8	3.6	2.12	0.044	2.26	1.02	4.38	307	0.45
15q11.2_del	osteoporosis	371396	1537	11626	33	48.1	0.69	0.060	0.73	0.50	1.01	363	0.52
16p11.2_dup	osteoporosis	369983	124	11600	7	3.9	1.76	0.083	2.10	0.90	4.21	463	0.56
22q11.2_dup	osteoporosis	370114	255	11604	11	8.0	1.36	0.10	1.73	0.89	3.04	529	0.60
PWS_dup	osteoporosis	369874	15	11594	1	0.5	2.06	0.14	5.08	0.51	24.00	662	0.65
13q12dup_CRYL1	osteoporosis	369868	9	11594	1	0.3	3.29	0.17	4.30	0.45	20.13	747	0.71
15q13.3_del	osteoporosis	369894	35	11595	2	1.1	1.78	0.18	2.81	0.56	8.80	774	0.72
13q12.12_dup	osteoporosis	370068	209	11596	3	6.5	0.47	0.20	0.54	0.15	1.34	858	0.74
1q21.1_del	osteoporosis	369960	101	11598	5	3.2	1.55	0.21	1.85	0.67	4.11	874	0.75
1q21.1_dup	osteoporosis	370025	166	11595	2	5.2	0.39	0.22	0.50	0.10	1.42	898	0.76
16p12.1_del	osteoporosis	370084	225	11603	10	7.1	1.40	0.22	1.52	0.76	2.72	909	0.77
22q11.2distal_dup	osteoporosis	369870	11	11594	1	0.3	2.74	0.24	3.41	0.35	15.95	951	0.78
2q13_del	osteoporosis	369908	49	11595	2	1.5	1.29	0.24	2.40	0.48	7.36	974	0.78
2q13_dup	osteoporosis	369930	71	11593	0	2.2	0.00	0.25	0.27	0.00	1.91	996	0.78
17p12_CMT1A_dup	osteoporosis	369970	111	11598	5	3.5	1.42	0.27	1.70	0.62	3.74	1056	0.80
16p11.2distal_dup	osteoporosis	369980	121	11599	6	3.8	1.55	0.30	1.64	0.60	3.62	1157	0.81
13q12.12_del	osteoporosis	369939	80	11597	4	2.5	1.57	0.30	1.74	0.56	4.15	1165	0.81
2q13dup(NPHP1)	osteoporosis	371664	1805	11656	63	56.6	1.11	0.31	1.14	0.88	1.46	1200	0.81
10q11.21q11.23_del	osteoporosis	369912	53	11596	3	1.7	1.76	0.33	1.83	0.49	4.90	1250	0.82
15q13.3_dup	osteoporosis	370080	221	11596	3	6.9	0.44	0.35	0.63	0.17	1.57	1324	0.84
2q21.1_dup	osteoporosis	369912	53	11595	2	1.7	1.20	0.36	1.95	0.40	5.93	1333	0.84
15q11q13dup_BP3_BP4	osteoporosis	369907	48	11595	2	1.5	1.32	0.36	1.95	0.39	6.11	1350	0.85
7q11.23_dup_distal	osteoporosis	369881	22	11594	1	0.7	1.43	0.37	2.37	0.26	9.82	1379	0.85
17p12_HNPP_del	osteoporosis	370069	210	11602	9	6.6	1.35	0.39	1.35	0.65	2.49	1460	0.85
17q12_del	osteoporosis	369866	7	11593	0	0.2	0.00	0.40	4.55	0.03	38.40	1470	0.85
10q23_dup	osteoporosis	369866	7	11593	0	0.2	0.00	0.41	4.47	0.03	42.18	1487	0.86
17q12_dup	osteoporosis	369955	96	11596	3	3.0	1.00	0.44	1.58	0.43	4.07	1581	0.87
22q11.2_del	osteoporosis	369869	10	11593	0	0.3	0.00	0.45	3.82	0.03	32.17	1602	0.87
2q21.1_del	osteoporosis	369899	40	11593	0	1.3	0.00	0.45	0.39	0.00	2.88	1604	0.87
3q29_dup	osteoporosis	369864	5	11593	0	0.16	0.00	0.45	4.12	0.03	72.24	1609	0.87
15q11.2_dup	osteoporosis	371723	1864	11646	53	58.4	0.91	0.48	0.91	0.68	1.18	1698	0.88
16p11.2distal_del	osteoporosis	369913	54	11593	0	1.7	0.00	0.50	0.44	0.00	3.10	1768	0.89
16p12.1_dup	osteoporosis	370044	185	11600	7	5.8	1.20	0.51	1.30	0.56	2.55	1776	0.89
8p23.1_dup	osteoporosis	369864	5	11593	0	0.2	0.00	0.51	3.14	0.02	32.24	1793	0.90
16p11.2_del	osteoporosis	369961	102	11594	1	3.2	0.32	0.58	0.65	0.07	2.42	1954	0.93
15q13.3del(CHRNA7)	osteoporosis	369869	10	11593	0	0.3	0.00	0.59	2.47	0.02	24.07	1998	0.93
2q11.2_dup	osteoporosis	369885	26	11593	0	0.8	0.00	0.65	0.55	0.00	4.19	2149	0.94
3q29_del	osteoporosis	369868	9	11593	0	0.3	0.00	0.66	2.06	0.02	18.06	2174	0.95
NRXN1_del	osteoporosis	370013	154	11596	3	4.8	0.63	0.67	0.80	0.22	2.02	2214	0.95
15q24_dup	osteoporosis	369867	8	11593	0	0.3	0.00	0.68	1.93	0.01	17.16	2253	0.95
TAR_dup	osteoporosis	370255	396	11605	12	12.4	0.97	0.69	1.12	0.60	1.91	2278	0.95
13q12del_CRYL1	osteoporosis	370212	353	11603	10	11.1	0.91	0.75	1.11	0.56	1.96	2447	0.96
TAR_del	osteoporosis	369927	68	11594	1	2.1	0.48	0.78	0.80	0.09	2.98	2521	0.97
17q11.2_del_NF1	osteoporosis	369868	9	11593	0	0.3	0.00	0.79	1.52	0.01	12.86	2556	0.97
15q11q13dup_BP3_BP5	osteoporosis	369868	9	11593	0	0.3	0.00	0.82	1.44	0.01	14.44	2646	0.97
WBS_dup	osteoporosis	369872	13	11593	0	0.4	0.00	0.83	1.40	0.01	11.61	2657	0.97
15q11q13del_BP3-BP4	osteoporosis	369873	14	11593	0	0.4	0.00	0.85	1.34	0.01	10.88	2730	0.97
2q11.2_del	osteoporosis	369888	29	11593	0	0.9	0.00	0.85	0.77	0.01	5.78	2743	0.97
16p13.11_dup	osteoporosis	370619	760	11616	23	23.8	0.97	0.94	1.02	0.65	1.51	2985	0.99
15q13.3dup(CHRNA7)	osteoporosis	372617	2758	11678	85	86.4	0.98	0.98	1.00	0.80	1.24	3083	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p12.1_del	ovarian_cysts	374288	224	7399	11	4.4	2.42	0.0070	2.58	1.33	4.52	95	0.23
15q11q13del_BP3-BP4	ovarian_cysts	374076	12	7390	2	0.2	7.38	0.0085	11.86	2.12	49.73	106	0.25
17q11.2_del_NF1	ovarian_cysts	374072	8	7389	1	0.2	5.74	0.032	13.67	1.32	83.50	255	0.40
13q12del_CRYL1	ovarian_cysts	374425	361	7390	2	7.1	0.28	0.038	0.34	0.07	0.95	276	0.43
2q11.2_dup	ovarian_cysts	374088	24	7390	2	0.5	3.97	0.042	5.54	1.08	18.61	299	0.44
16p11.2distal_del	ovarian_cysts	374115	51	7391	3	1.0	2.87	0.050	3.77	1.00	10.35	329	0.47
15q11.2_dup	ovarian_cysts	375954	1890	7415	27	37.3	0.73	0.070	0.72	0.48	1.03	401	0.54
16p13.11_del	ovarian_cysts	374188	124	7388	0	2.4	0.00	0.082	0.17	0.00	1.17	460	0.56
22q11.2distal_dup	ovarian_cysts	374075	11	7389	1	0.2	4.30	0.085	6.93	0.71	34.76	469	0.57
TAR_del	ovarian_cysts	374130	66	7391	3	1.3	2.24	0.13	2.62	0.71	6.89	639	0.64
22q11.2_dup	ovarian_cysts	374322	258	7396	8	5.1	1.55	0.13	1.79	0.82	3.38	640	0.64
1q21.1_del	ovarian_cysts	374170	106	7388	0	2.1	0.00	0.15	0.22	0.00	1.50	701	0.69
2q13_del	ovarian_cysts	374113	49	7390	2	1.0	2.02	0.16	2.99	0.60	9.36	706	0.69
17q12_dup	ovarian_cysts	374160	96	7391	3	1.9	1.56	0.16	2.45	0.66	6.44	713	0.69
2q13dup(NPHP1)	ovarian_cysts	375889	1825	7431	43	36.1	1.19	0.20	1.23	0.89	1.64	857	0.74
7q11.23_dup_distal	ovarian_cysts	374086	22	7389	1	0.4	2.24	0.25	3.20	0.35	13.27	990	0.78
3q29_dup	ovarian_cysts	374069	5	7388	0	0.10	0.00	0.28	8.99	0.06	170.61	1076	0.80
16p11.2distal_dup	ovarian_cysts	374187	123	7392	4	2.4	1.63	0.28	1.77	0.58	4.11	1101	0.80
1q21.1_dup	ovarian_cysts	374227	163	7393	5	3.2	1.54	0.29	1.65	0.61	3.56	1131	0.81
10q11.21q11.23_del	ovarian_cysts	374118	54	7390	2	1.1	1.84	0.31	2.11	0.43	6.36	1176	0.81
16p11.2_del	ovarian_cysts	374164	100	7391	3	2.0	1.50	0.32	1.82	0.50	4.71	1236	0.82
NRXN1_del	ovarian_cysts	374220	156	7389	1	3.1	0.33	0.33	0.49	0.06	1.77	1241	0.82
10q11.21q11.23_dup	ovarian_cysts	374103	39	7389	1	0.8	1.29	0.34	2.54	0.28	10.23	1272	0.83
8p23.1_dup	ovarian_cysts	374069	5	7388	0	0.1	0.00	0.35	5.95	0.04	73.34	1306	0.83
2q13_dup	ovarian_cysts	374133	69	7390	2	1.4	1.45	0.38	1.88	0.38	5.63	1402	0.85
TAR_dup	ovarian_cysts	374463	399	7397	9	7.9	1.14	0.43	1.32	0.64	2.39	1537	0.87
17q12_del	ovarian_cysts	374071	7	7388	0	0.1	0.00	0.43	4.23	0.03	52.32	1546	0.87
10q23_dup	ovarian_cysts	374071	7	7388	0	0.1	0.00	0.43	4.14	0.03	50.93	1560	0.87
Potocki_Lupski	ovarian_cysts	374069	5	7388	0	0.1	0.00	0.44	3.96	0.03	40.93	1585	0.87
16p11.2_dup	ovarian_cysts	374192	128	7391	3	2.5	1.18	0.46	1.54	0.42	3.93	1644	0.88
15q13.3del(CHRNA7)	ovarian_cysts	374074	10	7388	0	0.2	0.00	0.49	3.34	0.02	34.92	1737	0.89
16p13.11_dup	ovarian_cysts	374835	771	7400	12	15.2	0.79	0.50	0.83	0.45	1.39	1748	0.89
22q11.2distal_del	ovarian_cysts	374069	5	7388	0	0.1	0.00	0.51	3.11	0.02	32.29	1801	0.89
3q29_del	ovarian_cysts	374073	9	7388	0	0.2	0.00	0.55	2.80	0.02	26.31	1874	0.91
2q21.1_dup	ovarian_cysts	374118	54	7389	1	1.1	0.94	0.55	1.71	0.19	6.63	1897	0.92
15q13.3dup(CHRNA7)	ovarian_cysts	376856	2792	7439	51	55.1	0.93	0.56	0.92	0.69	1.20	1898	0.92
17p12_CMT1A_dup	ovarian_cysts	374179	115	7389	1	2.3	0.45	0.57	0.65	0.07	2.37	1940	0.92
15q11q13dup_BP3_BP5	ovarian_cysts	374073	9	7388	0	0.2	0.00	0.63	2.24	0.02	19.98	2078	0.94
22q11.2_del	ovarian_cysts	374074	10	7388	0	0.2	0.00	0.63	2.20	0.02	20.76	2110	0.94
15q13.3_dup	ovarian_cysts	374286	222	7390	2	4.4	0.46	0.66	0.76	0.16	2.19	2170	0.95
13q12.12_dup	ovarian_cysts	374273	209	7391	3	4.1	0.73	0.68	0.80	0.22	2.01	2224	0.95
15q24_dup	ovarian_cysts	374072	8	7388	0	0.2	0.00	0.69	1.91	0.01	16.30	2259	0.95
13q12dup_CRYL1	ovarian_cysts	374074	10	7388	0	0.2	0.00	0.71	1.82	0.01	15.01	2308	0.96
2q21.1_del	ovarian_cysts	374104	40	7388	0	0.8	0.00	0.72	0.62	0.00	4.50	2329	0.96
WBS_dup	ovarian_cysts	374077	13	7388	0	0.3	0.00	0.72	1.76	0.01	14.95	2358	0.96
16p12.1_dup	ovarian_cysts	374253	189	7391	3	3.7	0.81	0.76	0.85	0.24	2.12	2456	0.96
15q11.2_del	ovarian_cysts	375604	1540	7418	30	30.4	0.99	0.76	1.06	0.72	1.49	2468	0.97
15q13.3_del	ovarian_cysts	374101	37	7388	0	0.7	0.00	0.77	0.68	0.01	4.98	2495	0.97
15q11q13dup_BP3_BP4	ovarian_cysts	374114	50	7388	0	1.0	0.00	0.78	0.68	0.01	4.98	2515	0.97
17p12_HNPP_del	ovarian_cysts	374280	216	7391	3	4.3	0.71	0.79	0.87	0.24	2.17	2549	0.97
2q13del(NPHP1)	ovarian_cysts	376350	2286	7432	44	45.1	0.98	0.81	0.96	0.70	1.28	2606	0.97
13q12.12_del	ovarian_cysts	374147	83	7389	1	1.6	0.61	0.88	0.88	0.10	3.27	2805	0.98
PWS_dup	ovarian_cysts	374080	16	7388	0	0.3	0.00	0.99	0.99	0.01	7.72	3119	1.00
2q11.2_del	ovarian_cysts	374093	29	7388	0	0.6	0.00	1.00	1.01	0.01	7.67	3122	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
22q11.2distal_del	paralytic_syndromes	379496	4	1961	1	0.0	38.92	0.0038	51.58	5.07	289.93	71	0.17
17q12_del	paralytic_syndromes	379498	6	1961	1	0.0	27.80	0.0083	30.14	3.09	148.20	103	0.25
15q11q13del_BP3-BP4	paralytic_syndromes	379505	13	1961	1	0.1	13.90	0.012	22.33	2.41	94.04	138	0.28
22q11.2distal_dup	paralytic_syndromes	379503	11	1961	1	0.1	16.22	0.015	20.10	2.16	85.60	154	0.30
PWS_dup	paralytic_syndromes	379507	15	1961	1	0.1	12.16	0.015	19.39	2.11	79.69	158	0.30
10q11.21q11.23_del	paralytic_syndromes	379546	54	1962	2	0.3	6.95	0.015	7.91	1.63	23.27	159	0.30
13q12del_CRYL1	paralytic_syndromes	379850	358	1965	5	1.9	2.68	0.023	3.18	1.20	6.71	208	0.35
16p11.2_dup	paralytic_syndromes	379620	128	1963	3	0.7	4.46	0.025	4.49	1.25	11.26	219	0.36
17q12_dup	paralytic_syndromes	379589	97	1962	2	0.5	3.93	0.063	4.35	0.90	12.53	372	0.53
2q21.1_del	paralytic_syndromes	379531	39	1961	1	0.2	4.87	0.078	6.68	0.75	25.12	444	0.55
16p13.11_del	paralytic_syndromes	379614	122	1962	2	0.6	3.14	0.079	3.95	0.82	11.30	447	0.55
17p12_CMT1A_dup	paralytic_syndromes	379606	114	1962	2	0.6	3.36	0.081	3.89	0.81	11.18	458	0.56
16p12.1_del	paralytic_syndromes	379724	232	1963	3	1.2	2.48	0.11	2.69	0.75	6.66	569	0.63
2q13_del	paralytic_syndromes	379542	50	1961	1	0.3	3.82	0.12	5.01	0.56	18.63	606	0.63
16p11.2distal_del	paralytic_syndromes	379545	53	1961	1	0.3	3.60	0.14	4.66	0.53	17.22	654	0.65
3q29_dup	paralytic_syndromes	379497	5	1960	0	0.03	0.00	0.16	18.57	0.14	168.98	735	0.70
8p23.1_dup	paralytic_syndromes	379497	5	1960	0	0.0	0.00	0.18	16.75	0.13	150.07	763	0.72
Potocki_Lupski	paralytic_syndromes	379497	5	1960	0	0.0	0.00	0.19	15.36	0.12	139.87	799	0.73
1q21.1_dup	paralytic_syndromes	379658	166	1962	2	0.9	2.32	0.20	2.58	0.54	7.35	830	0.75
15q24_dup	paralytic_syndromes	379500	8	1960	0	0.0	0.00	0.21	12.36	0.09	102.91	879	0.75
13q12.12_del	paralytic_syndromes	379575	83	1961	1	0.4	2.32	0.21	3.42	0.39	12.46	886	0.76
10q23_dup	paralytic_syndromes	379499	7	1960	0	0.0	0.00	0.23	10.91	0.08	93.48	930	0.77
13q12dup_CRYL1	paralytic_syndromes	379502	10	1960	0	0.1	0.00	0.24	10.15	0.08	79.83	954	0.78
17q11.2_del_NF1	paralytic_syndromes	379501	9	1960	0	0.0	0.00	0.25	9.50	0.07	75.82	998	0.78
15q11q13dup_BP3_BP5	paralytic_syndromes	379501	9	1960	0	0.0	0.00	0.26	9.08	0.07	73.83	1020	0.79
3q29_del	paralytic_syndromes	379501	9	1960	0	0.0	0.00	0.27	8.22	0.06	65.71	1063	0.80
15q11.2_del	paralytic_syndromes	381057	1565	1965	5	8.1	0.62	0.28	0.65	0.25	1.36	1085	0.80
22q11.2_del	paralytic_syndromes	379502	10	1960	0	0.1	0.00	0.31	6.70	0.05	53.99	1193	0.81
15q13.3del(CHRNA7)	paralytic_syndromes	379502	10	1960	0	0.1	0.00	0.32	6.52	0.05	52.45	1214	0.81
WBS_dup	paralytic_syndromes	379505	13	1960	0	0.1	0.00	0.34	5.85	0.05	44.66	1275	0.83
7q11.23_dup_distal	paralytic_syndromes	379515	23	1960	0	0.1	0.00	0.40	4.51	0.04	32.79	1463	0.85
16p11.2distal_dup	paralytic_syndromes	379618	126	1961	1	0.7	1.53	0.40	2.17	0.25	7.83	1481	0.85
TAR_dup	paralytic_syndromes	379897	405	1963	3	2.1	1.43	0.42	1.59	0.45	3.93	1521	0.87
2q11.2_dup	paralytic_syndromes	379518	26	1960	0	0.1	0.00	0.44	3.89	0.03	27.98	1563	0.87
2q11.2_del	paralytic_syndromes	379521	29	1960	0	0.1	0.00	0.56	2.63	0.02	18.95	1909	0.91
10q11.21q11.23_dup	paralytic_syndromes	379532	40	1960	0	0.2	0.00	0.60	2.36	0.02	16.64	2006	0.93
15q13.3dup(CHRNA7)	paralytic_syndromes	382323	2831	1972	12	14.6	0.82	0.61	0.87	0.47	1.45	2023	0.94
16p12.1_dup	paralytic_syndromes	379683	191	1961	1	1.0	1.01	0.64	1.51	0.17	5.43	2115	0.94
2q13del(NPHP1)	paralytic_syndromes	381812	2320	1970	10	12.0	0.84	0.67	0.88	0.45	1.53	2193	0.95
15q11.2_dup	paralytic_syndromes	381398	1906	1971	11	9.8	1.12	0.67	1.14	0.60	1.93	2219	0.95
NRXN1_del	paralytic_syndromes	379649	157	1960	0	0.8	0.00	0.68	0.59	0.00	4.02	2244	0.95
15q13.3_del	paralytic_syndromes	379529	37	1960	0	0.2	0.00	0.71	1.77	0.01	12.53	2322	0.96
15q11q13dup_BP3_BP4	paralytic_syndromes	379542	50	1960	0	0.3	0.00	0.72	1.73	0.01	12.10	2356	0.96
13q12.12_dup	paralytic_syndromes	379703	211	1961	1	1.1	0.92	0.73	1.35	0.15	4.83	2379	0.96
17p12_HNPP_del	paralytic_syndromes	379710	218	1961	1	1.1	0.89	0.75	1.32	0.15	4.73	2419	0.97
16p11.2_del	paralytic_syndromes	379595	103	1960	0	0.5	0.00	0.75	0.66	0.01	4.52	2430	0.97
2q21.1_dup	paralytic_syndromes	379547	55	1960	0	0.3	0.00	0.77	1.57	0.01	10.95	2489	0.97
TAR_del	paralytic_syndromes	379561	69	1960	0	0.4	0.00	0.81	1.42	0.01	9.89	2623	0.97
2q13_dup	paralytic_syndromes	379563	71	1960	0	0.4	0.00	0.85	1.33	0.01	9.19	2733	0.97
16p13.11_dup	paralytic_syndromes	380271	779	1964	4	4.0	0.99	0.89	1.07	0.36	2.40	2825	0.98
1q21.1_del	paralytic_syndromes	379598	106	1960	0	0.5	0.00	0.92	0.87	0.01	5.95	2918	0.98
15q13.3_dup	paralytic_syndromes	379715	223	1961	1	1.2	0.87	0.92	1.09	0.12	3.90	2928	0.98
22q11.2_dup	paralytic_syndromes	379757	265	1961	1	1.4	0.73	0.93	0.93	0.11	3.34	2957	0.99
2q13dup(NPHP1)	paralytic_syndromes	381351	1859	1969	9	9.6	0.94	0.98	0.99	0.48	1.77	3077	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
17q12_dup	prostate_hyperplasia	367092	87	14459	12	3.4	3.20	0.0044	2.84	1.41	5.33	75	0.19
16p13.11_dup	prostate_hyperplasia	367748	743	14487	40	29.3	1.35	0.011	1.58	1.11	2.19	130	0.27
15q11.2_del	prostate_hyperplasia	368528	1523	14494	47	59.9	0.79	0.014	0.70	0.51	0.93	151	0.30
22q11.2distal_dup	prostate_hyperplasia	367015	10	14449	2	0.4	4.40	0.039	7.01	1.12	35.12	283	0.43
15q13.3_dup	prostate_hyperplasia	367225	220	14451	4	8.7	0.47	0.040	0.42	0.14	0.97	294	0.43
17p12_CMT1A_dup	prostate_hyperplasia	367113	108	14455	8	4.3	1.82	0.046	2.34	1.02	4.82	315	0.45
2q13_del	prostate_hyperplasia	367052	47	14451	4	1.9	2.07	0.087	2.80	0.84	7.52	480	0.57
15q24_dup	prostate_hyperplasia	367013	8	14447	0	0.3	0.00	0.12	30.95	0.22	383.60	613	0.63
13q12dup_CRYL1	prostate_hyperplasia	367014	9	14448	1	0.4	2.64	0.13	6.78	0.52	76.51	632	0.64
15q11.2_dup	prostate_hyperplasia	368840	1835	14529	82	72.3	1.13	0.13	1.20	0.95	1.51	636	0.64
15q11q13del_BP3-BP4	prostate_hyperplasia	367018	13	14448	1	0.5	1.89	0.14	5.10	0.50	27.90	675	0.67
22q11.2_del	prostate_hyperplasia	367014	9	14448	1	0.4	2.64	0.15	5.10	0.48	30.26	689	0.68
10q23_dup	prostate_hyperplasia	367011	6	14448	1	0.2	3.77	0.17	5.07	0.44	38.58	744	0.71
13q12del_CRYL1	prostate_hyperplasia	367360	355	14455	8	14.0	0.58	0.18	0.64	0.29	1.21	770	0.72
15q13.3_del	prostate_hyperplasia	367039	34	14450	3	1.3	2.14	0.20	2.32	0.59	6.86	849	0.74
16p11.2_del	prostate_hyperplasia	367107	102	14448	1	4.0	0.26	0.22	0.41	0.05	1.55	907	0.77
16p12.1_del	prostate_hyperplasia	367229	224	14458	11	8.8	1.24	0.24	1.49	0.75	2.70	971	0.78
2q21.1_del	prostate_hyperplasia	367042	37	14450	3	1.5	1.98	0.24	2.13	0.55	6.24	980	0.78
10q11.21q11.23_dup	prostate_hyperplasia	367043	38	14449	2	1.5	1.32	0.25	2.42	0.47	8.06	1007	0.78
1q21.1_del	prostate_hyperplasia	367106	101	14452	5	4.0	1.25	0.27	1.74	0.61	4.06	1062	0.80
16p12.1_dup	prostate_hyperplasia	367193	188	14451	4	7.4	0.55	0.28	0.61	0.20	1.44	1103	0.80
1q21.1_dup	prostate_hyperplasia	367165	160	14455	8	6.3	1.26	0.30	1.51	0.67	3.00	1156	0.81
WBS_dup	prostate_hyperplasia	367017	12	14448	1	0.5	2.03	0.33	2.85	0.27	16.95	1248	0.82
TAR_del	prostate_hyperplasia	367070	65	14451	4	2.6	1.53	0.33	1.73	0.53	4.48	1252	0.82
TAR_dup	prostate_hyperplasia	367401	396	14459	12	15.6	0.78	0.34	0.76	0.40	1.31	1292	0.83
16p11.2_dup	prostate_hyperplasia	367133	128	14450	3	5.0	0.60	0.35	0.61	0.17	1.59	1305	0.83
22q11.2distal_del	prostate_hyperplasia	367010	5	14447	0	0.2	0.00	0.39	5.05	0.04	72.86	1445	0.85
16p13.11_del	prostate_hyperplasia	367124	119	14452	5	4.7	1.06	0.42	1.48	0.53	3.46	1527	0.87
2q11.2_del	prostate_hyperplasia	367034	29	14447	0	1.1	0.00	0.44	0.38	0.00	2.97	1562	0.87
3q29_dup	prostate_hyperplasia	367010	5	14447	0	0.20	0.00	0.44	4.08	0.03	44.96	1569	0.87
2q21.1_dup	prostate_hyperplasia	367059	54	14448	1	2.1	0.48	0.44	0.55	0.06	2.15	1589	0.87
17q11.2_del_NF1	prostate_hyperplasia	367013	8	14448	1	0.3	2.93	0.44	2.22	0.21	13.28	1596	0.87
16p11.2distal_dup	prostate_hyperplasia	367130	125	14449	2	4.9	0.42	0.44	0.63	0.13	1.87	1598	0.87
2q13_dup	prostate_hyperplasia	367072	67	14451	4	2.6	1.49	0.45	1.50	0.47	3.80	1619	0.88
2q13del(NPHP1)	prostate_hyperplasia	369241	2236	14541	94	88.1	1.07	0.46	1.09	0.87	1.34	1635	0.88
NRXN1_del	prostate_hyperplasia	367158	153	14451	4	6.0	0.67	0.48	0.71	0.23	1.70	1694	0.88
2q11.2_dup	prostate_hyperplasia	367030	25	14448	1	1.0	1.02	0.49	1.97	0.20	9.50	1731	0.89
15q11q13dup_BP3_BP5	prostate_hyperplasia	367014	9	14447	0	0.4	0.00	0.52	3.08	0.02	37.19	1827	0.90
7q11.23_dup_distal	prostate_hyperplasia	367027	22	14448	1	0.9	1.15	0.57	1.76	0.18	8.81	1933	0.92
15q13.3dup(CHRNA7)	prostate_hyperplasia	369748	2743	14547	100	107.9	0.93	0.59	0.94	0.76	1.16	1993	0.93
10q11.21q11.23_del	prostate_hyperplasia	367059	54	14449	2	2.1	0.94	0.61	1.44	0.28	4.75	2050	0.94
17p12_HNPP_del	prostate_hyperplasia	367215	210	14456	9	8.3	1.09	0.74	1.12	0.53	2.12	2413	0.97
Potocki_Lupski	prostate_hyperplasia	367010	5	14447	0	0.2	0.00	0.75	1.73	0.01	28.01	2423	0.97
13q12.12_del	prostate_hyperplasia	367086	81	14450	3	3.2	0.94	0.76	0.84	0.23	2.28	2469	0.97
15q11q13dup_BP3_BP4	prostate_hyperplasia	367054	49	14448	1	1.9	0.53	0.77	0.78	0.09	3.16	2483	0.97
13q12.12_dup	prostate_hyperplasia	367211	206	14453	6	8.1	0.75	0.79	0.90	0.36	1.88	2548	0.97
2q13dup(NPHP1)	prostate_hyperplasia	368797	1792	14523	76	70.6	1.07	0.83	1.03	0.80	1.30	2669	0.97
15q13.3del(CHRNA7)	prostate_hyperplasia	367015	10	14447	0	0.4	0.00	0.86	1.32	0.01	11.36	2759	0.97
22q11.2_dup	prostate_hyperplasia	367262	257	14456	9	10.1	0.89	0.90	1.04	0.50	1.95	2876	0.98
16p11.2distal_del	prostate_hyperplasia	367057	52	14449	2	2.0	0.98	0.94	1.05	0.21	3.28	2983	0.99
3q29_del	prostate_hyperplasia	367014	9	14447	0	0.4	0.00	0.94	1.12	0.01	11.66	2989	0.99
8p23.1_dup	prostate_hyperplasia	367010	5	14447	0	0.2	0.00	0.94	1.12	0.01	13.22	2991	0.99
PWS_dup	prostate_hyperplasia	367021	16	14447	0	0.6	0.00	0.94	0.90	0.01	10.68	2997	0.99
17q12_del	prostate_hyperplasia	367012	7	14447	0	0.3	0.00	0.96	0.93	0.01	8.50	3034	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2_del	psoriasis	375530	98	6025	5	1.6	3.08	0.039	2.86	1.06	6.16	285	0.43
2q13dup(NPHP1)	psoriasis	377280	1848	6040	20	29.6	0.68	0.077	0.69	0.43	1.04	436	0.55
2q13_dup	psoriasis	375500	68	6023	3	1.1	2.68	0.077	3.12	0.86	7.91	437	0.55
16p12.1_dup	psoriasis	375618	186	6026	6	3.0	1.98	0.080	2.20	0.90	4.42	451	0.55
13q12del_CRYL1	psoriasis	375786	354	6029	9	5.7	1.57	0.12	1.75	0.85	3.14	593	0.63
16p11.2_dup	psoriasis	375563	131	6020	0	2.1	0.00	0.16	0.22	0.00	1.53	729	0.70
10q11.21q11.23_del	psoriasis	375486	54	6022	2	0.9	2.26	0.18	2.74	0.57	8.01	772	0.72
2q11.2_dup	psoriasis	375457	25	6021	1	0.4	2.44	0.19	3.74	0.42	14.36	811	0.74
16p13.11_del	psoriasis	375556	124	6020	0	2.0	0.00	0.21	0.25	0.00	1.74	885	0.76
17q12_dup	psoriasis	375528	96	6023	3	1.5	1.92	0.22	2.10	0.58	5.28	906	0.77
TAR_dup	psoriasis	375831	399	6029	9	6.4	1.40	0.27	1.46	0.71	2.63	1070	0.80
22q11.2_dup	psoriasis	375696	264	6022	2	4.2	0.48	0.30	0.55	0.12	1.56	1173	0.81
8p23.1_dup	psoriasis	375437	5	6020	0	0.1	0.00	0.34	5.83	0.04	51.56	1294	0.83
3q29_dup	psoriasis	375437	5	6020	0	0.08	0.00	0.36	5.39	0.04	48.12	1338	0.84
16p11.2distal_dup	psoriasis	375556	124	6023	3	2.0	1.50	0.36	1.71	0.48	4.26	1345	0.85
Potocki_Lupski	psoriasis	375437	5	6020	0	0.1	0.00	0.37	5.16	0.04	46.17	1369	0.85
22q11.2distal_del	psoriasis	375437	5	6020	0	0.1	0.00	0.38	5.02	0.04	44.39	1389	0.85
2q21.1_del	psoriasis	375471	39	6021	1	0.6	1.58	0.38	2.28	0.26	8.52	1403	0.85
13q12.12_del	psoriasis	375514	82	6022	2	1.3	1.51	0.38	1.86	0.39	5.35	1404	0.85
15q13.3_del	psoriasis	375468	36	6021	1	0.6	1.71	0.40	2.20	0.25	8.25	1477	0.85
10q23_dup	psoriasis	375439	7	6020	0	0.1	0.00	0.43	4.06	0.03	33.45	1541	0.87
15q13.3_dup	psoriasis	375651	219	6025	5	3.5	1.41	0.45	1.41	0.53	2.99	1608	0.87
15q24_dup	psoriasis	375440	8	6020	0	0.1	0.00	0.46	3.56	0.03	28.86	1652	0.88
17q12_del	psoriasis	375439	7	6020	0	0.1	0.00	0.47	3.55	0.03	29.39	1657	0.88
16p12.1_del	psoriasis	375665	233	6022	2	3.7	0.54	0.48	0.66	0.14	1.85	1687	0.88
NRXN1_del	psoriasis	375588	156	6021	1	2.5	0.40	0.48	0.59	0.07	2.12	1705	0.88
17q11.2_del_NF1	psoriasis	375441	9	6020	0	0.1	0.00	0.50	3.17	0.02	25.07	1752	0.89
3q29_del	psoriasis	375441	9	6020	0	0.1	0.00	0.51	3.06	0.02	24.17	1787	0.89
15q11q13dup_BP3_BP5	psoriasis	375441	9	6020	0	0.1	0.00	0.53	2.88	0.02	22.88	1842	0.90
13q12dup_CRYL1	psoriasis	375442	10	6020	0	0.2	0.00	0.54	2.78	0.02	21.68	1866	0.91
2q13_del	psoriasis	375482	50	6021	1	0.8	1.24	0.56	1.70	0.19	6.27	1901	0.92
2q21.1_dup	psoriasis	375486	54	6021	1	0.9	1.15	0.57	1.65	0.19	6.09	1943	0.92
22q11.2_del	psoriasis	375442	10	6020	0	0.2	0.00	0.58	2.48	0.02	19.42	1960	0.93
15q13.3del(CHRNA7)	psoriasis	375442	10	6020	0	0.2	0.00	0.59	2.45	0.02	19.20	1974	0.93
13q12.12_dup	psoriasis	375642	210	6022	2	3.4	0.60	0.60	0.73	0.15	2.06	2010	0.93
16p11.2distal_del	psoriasis	375486	54	6020	0	0.9	0.00	0.61	0.52	0.00	3.63	2039	0.94
22q11.2distal_dup	psoriasis	375444	12	6020	0	0.2	0.00	0.61	2.28	0.02	17.39	2048	0.94
WBS_dup	psoriasis	375445	13	6020	0	0.2	0.00	0.63	2.17	0.02	16.42	2096	0.94
15q11q13del_BP3-BP4	psoriasis	375446	14	6020	0	0.2	0.00	0.63	2.16	0.02	16.23	2105	0.94
15q11q13dup_BP3_BP4	psoriasis	375482	50	6020	0	0.8	0.00	0.66	0.57	0.00	3.95	2180	0.95
1q21.1_dup	psoriasis	375597	165	6023	3	2.6	1.13	0.68	1.26	0.35	3.13	2226	0.95
PWS_dup	psoriasis	375448	16	6020	0	0.3	0.00	0.68	1.90	0.01	14.06	2254	0.95
TAR_del	psoriasis	375500	68	6021	1	1.1	0.92	0.71	1.37	0.16	5.02	2323	0.96
2q13del(NPHP1)	psoriasis	377724	2292	6058	38	36.8	1.03	0.73	1.06	0.75	1.43	2393	0.96
17p12_CMT1A_dup	psoriasis	375547	115	6021	1	1.8	0.55	0.77	0.80	0.09	2.88	2505	0.97
10q11.21q11.23_dup	psoriasis	375472	40	6020	0	0.6	0.00	0.82	0.73	0.01	5.14	2634	0.97
7q11.23_dup_distal	psoriasis	375455	23	6020	0	0.4	0.00	0.82	1.41	0.01	10.16	2642	0.97
1q21.1_del	psoriasis	375537	105	6021	1	1.7	0.60	0.86	0.87	0.10	3.15	2772	0.98
15q13.3dup(CHRNA7)	psoriasis	378232	2800	6063	43	44.9	0.96	0.88	0.98	0.71	1.30	2822	0.98
15q11.2_del	psoriasis	376977	1545	6045	25	24.8	1.01	0.92	1.02	0.67	1.48	2923	0.98
2q11.2_del	psoriasis	375461	29	6020	0	0.5	0.00	0.96	0.93	0.01	6.63	3036	0.99
16p13.11_dup	psoriasis	376203	771	6032	12	12.4	0.97	0.97	0.99	0.53	1.65	3053	0.99
15q11.2_dup	psoriasis	377319	1887	6050	30	30.3	0.99	0.98	1.00	0.68	1.40	3087	1.00
17p12_HNPP_del	psoriasis	375648	216	6023	3	3.5	0.87	1.00	1.00	0.28	2.48	3121	1.00



CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
15q13.3_dup	renal	374375	212	7301	12	4.1	2.80	0.0017	2.89	1.55	4.91	49	0.11
17q12_del	renal	374168	5	7291	2	0.1	14.95	0.0021	20.82	3.71	87.49	57	0.11
16p11.2_del	renal	374260	97	7295	6	1.9	3.05	0.015	3.16	1.28	6.49	160	0.30
22q11.2distal_del	renal	374167	4	7290	1	0.1	10.47	0.023	16.79	1.66	92.62	204	0.35
17q12_dup	renal	374257	94	7294	5	1.8	2.64	0.035	2.93	1.09	6.31	268	0.41
2q13_del	renal	374211	48	7292	3	0.9	3.08	0.047	3.72	1.02	9.60	319	0.46
15q11q13dup_BP3_BP5	renal	374171	8	7290	1	0.2	5.81	0.053	9.12	0.97	41.02	341	0.49
3q29_del	renal	374171	8	7290	1	0.2	5.81	0.058	8.56	0.91	38.25	359	0.51
PWS_dup	renal	374178	15	7290	1	0.3	3.27	0.11	5.56	0.61	22.46	546	0.62
2q21.1_del	renal	374201	38	7291	2	0.7	2.62	0.13	3.21	0.66	9.56	631	0.64
2q11.2_dup	renal	374188	25	7290	1	0.5	2.01	0.25	3.12	0.35	11.97	1001	0.78
15q13.3dup(CHRNA7)	renal	376944	2781	7351	62	54.2	1.14	0.26	1.16	0.89	1.48	1018	0.79
13q12.12_del	renal	374247	84	7289	0	1.6	0.00	0.28	0.29	0.00	2.02	1099	0.80
16p11.2_dup	renal	374290	127	7293	4	2.5	1.60	0.29	1.74	0.58	3.96	1126	0.81
1q21.1_del	renal	374266	103	7292	3	2.0	1.48	0.34	1.75	0.49	4.39	1304	0.83
3q29_dup	renal	374168	5	7289	0	0.10	0.00	0.36	5.48	0.04	49.40	1332	0.84
2q13_dup	renal	374234	71	7289	0	1.4	0.00	0.40	0.37	0.00	2.52	1464	0.85
NRXN1_del	renal	374316	153	7293	4	3.0	1.33	0.40	1.54	0.51	3.49	1478	0.85
Potocki_Lupski	renal	374168	5	7289	0	0.1	0.00	0.40	4.48	0.03	39.68	1482	0.85
8p23.1_dup	renal	374168	5	7289	0	0.1	0.00	0.41	4.35	0.03	38.40	1497	0.86
22q11.2_dup	renal	374426	263	7292	3	5.1	0.59	0.42	0.67	0.19	1.64	1510	0.87
TAR_del	renal	374232	69	7289	0	1.3	0.00	0.42	0.38	0.00	2.63	1522	0.87
17p12_CMT1A_dup	renal	374276	113	7292	3	2.2	1.35	0.45	1.55	0.43	3.88	1613	0.87
2q13dup(NPHP1)	renal	376000	1837	7320	31	35.8	0.87	0.46	0.88	0.60	1.23	1641	0.88
10q23_dup	renal	374170	7	7289	0	0.1	0.00	0.47	3.54	0.03	29.69	1663	0.88
15q24_dup	renal	374171	8	7289	0	0.2	0.00	0.48	3.43	0.03	27.70	1688	0.88
16p13.11_del	renal	374284	121	7292	3	2.4	1.27	0.50	1.48	0.41	3.69	1751	0.89
15q13.3_del	renal	374199	36	7290	1	0.7	1.41	0.51	1.83	0.21	6.87	1779	0.89
16p11.2distal_dup	renal	374289	126	7290	1	2.5	0.41	0.53	0.62	0.07	2.24	1844	0.90
2q13del(NPHP1)	renal	376445	2282	7337	48	44.5	1.08	0.54	1.09	0.81	1.44	1871	0.91
17q11.2_del_NF1	renal	374172	9	7289	0	0.2	0.00	0.58	2.48	0.02	19.59	1964	0.93
13q12dup_CRYL1	renal	374173	10	7289	0	0.2	0.00	0.59	2.42	0.02	18.80	1991	0.93
15q11q13dup_BP3_BP4	renal	374213	50	7289	0	1.0	0.00	0.59	0.51	0.00	3.54	1994	0.93
22q11.2_del	renal	374173	10	7289	0	0.2	0.00	0.60	2.39	0.02	18.82	2000	0.93
15q13.3del(CHRNA7)	renal	374173	10	7289	0	0.2	0.00	0.61	2.27	0.02	17.78	2055	0.94
1q21.1_dup	renal	374329	166	7291	2	3.2	0.62	0.63	0.74	0.16	2.11	2086	0.94
16p12.1_del	renal	374393	230	7294	5	4.5	1.11	0.69	1.20	0.45	2.53	2258	0.95
15q11q13del_BP3-BP4	renal	374177	14	7289	0	0.3	0.00	0.69	1.88	0.01	14.10	2269	0.95
16p11.2distal_del	renal	374216	53	7290	1	1.0	0.97	0.69	1.41	0.16	5.19	2277	0.96
WBS_dup	renal	374176	13	7289	0	0.3	0.00	0.70	1.83	0.01	13.90	2294	0.96
2q21.1_dup	renal	374217	54	7290	1	1.1	0.95	0.71	1.38	0.16	5.09	2310	0.96
22q11.2distal_dup	renal	374175	12	7289	0	0.2	0.00	0.72	1.76	0.01	13.44	2340	0.96
16p13.11_dup	renal	374930	767	7305	16	14.9	1.07	0.72	1.09	0.64	1.72	2345	0.96
15q11.2_del	renal	375705	1542	7317	28	30.0	0.93	0.74	0.94	0.63	1.34	2416	0.97
10q11.21q11.23_del	renal	374218	55	7290	1	1.1	0.93	0.77	1.29	0.15	4.74	2490	0.97
10q11.21q11.23_dup	renal	374203	40	7289	0	0.8	0.00	0.77	0.68	0.01	4.78	2508	0.97
15q11.2_dup	renal	376042	1879	7327	38	36.6	1.04	0.80	1.04	0.74	1.42	2581	0.97
TAR_dup	renal	374563	400	7297	8	7.8	1.03	0.81	1.09	0.51	2.01	2605	0.97
13q12.12_dup	renal	374371	208	7293	4	4.1	0.99	0.81	1.12	0.37	2.54	2612	0.97
2q11.2_del	renal	374192	29	7289	0	0.6	0.00	0.89	0.83	0.01	5.92	2860	0.98
17p12_HNPP_del	renal	374378	215	7293	4	4.2	0.96	0.90	1.07	0.36	2.41	2866	0.98
7q11.23_dup_distal	renal	374186	23	7289	0	0.4	0.00	0.92	1.15	0.01	8.29	2935	0.99
16p12.1_dup	renal	374352	189	7292	3	3.7	0.82	0.93	0.95	0.27	2.36	2948	0.98
13q12del_CRYL1	renal	374520	357	7295	6	7.0	0.87	0.96	0.98	0.40	1.96	3042	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p12.1_del	renal_failure	373750	218	7937	17	4.6	3.48	7.23E-06	3.85	2.26	6.13	11	0.0021
16p11.2_del	renal_failure	373625	93	7930	10	2.0	4.68	6.04E-05	5.11	2.49	9.45	16	0.012
3q29_dup	renal_failure	373535	3	7922	2	0.06	19.27	0.00022	58.45	9.22	324.78	25	0.027
17q12_dup	renal_failure	373622	90	7929	9	1.9	4.38	0.00029	4.56	2.16	8.57	26	0.035
22q11.2distal_del	renal_failure	373536	4	7921	1	0.1	9.63	0.021	18.72	1.75	114.67	189	0.35
16p11.2_dup	renal_failure	373656	124	7927	7	2.6	2.57	0.025	2.65	1.15	5.23	216	0.36
17p12_CMT1A_dup	renal_failure	373642	110	7926	6	2.3	2.49	0.035	2.69	1.08	5.55	263	0.41
17q12_del	renal_failure	373538	6	7921	1	0.1	6.88	0.061	8.69	0.88	44.55	368	0.52
3q29_del	renal_failure	373540	8	7921	1	0.2	5.35	0.069	7.87	0.82	36.81	394	0.54
22q11.2_del	renal_failure	373541	9	7921	1	0.2	4.82	0.069	7.98	0.81	38.28	396	0.54
16p11.2distal_del	renal_failure	373583	51	7923	3	1.1	2.68	0.071	3.26	0.89	8.48	409	0.54
15q13.3_dup	renal_failure	373747	215	7929	9	4.6	1.94	0.085	1.88	0.91	3.43	471	0.57
2q13del(NPHP1)	renal_failure	375803	2271	7979	59	48.2	1.22	0.11	1.24	0.95	1.60	566	0.63
17p12_HNPP_del	renal_failure	373743	211	7928	8	4.5	1.76	0.12	1.83	0.84	3.43	580	0.63
13q12.12_dup	renal_failure	373743	211	7921	1	4.5	0.23	0.12	0.35	0.04	1.25	610	0.63
TAR_del	renal_failure	373598	66	7923	3	1.4	2.09	0.13	2.58	0.70	6.68	653	0.65
22q11.2_dup	renal_failure	373789	257	7929	9	5.5	1.63	0.14	1.69	0.82	3.08	676	0.67
16p13.11_dup	renal_failure	374293	761	7942	22	16.1	1.35	0.15	1.39	0.88	2.07	678	0.67
2q13_dup	renal_failure	373600	68	7923	3	1.4	2.04	0.16	2.42	0.66	6.25	719	0.69
15q13.3_del	renal_failure	373567	35	7922	2	0.7	2.60	0.20	2.64	0.53	8.10	832	0.75
7q11.23_dup_distal	renal_failure	373554	22	7921	1	0.5	2.09	0.21	3.50	0.38	13.98	888	0.76
NRXN1_del	renal_failure	373684	152	7925	5	3.2	1.53	0.23	1.76	0.66	3.78	945	0.78
16p13.11_del	renal_failure	373652	120	7924	4	2.5	1.55	0.24	1.86	0.61	4.30	967	0.78
13q12.12_del	renal_failure	373613	81	7923	3	1.7	1.72	0.29	1.90	0.52	4.85	1119	0.80
2q21.1_dup	renal_failure	373585	53	7922	2	1.1	1.75	0.31	2.10	0.43	6.25	1182	0.81
2q13dup(NPHP1)	renal_failure	375355	1823	7965	45	38.7	1.16	0.32	1.17	0.85	1.55	1235	0.82
15q11.2_del	renal_failure	375063	1531	7959	39	32.5	1.20	0.34	1.17	0.84	1.59	1297	0.83
1q21.1_del	renal_failure	373635	103	7923	3	2.2	1.36	0.37	1.70	0.47	4.31	1370	0.85
15q24_dup	renal_failure	373540	8	7920	0	0.2	0.00	0.38	4.97	0.04	41.16	1392	0.85
10q11.21q11.23_dup	renal_failure	373571	39	7921	1	0.8	1.20	0.41	2.17	0.24	8.32	1494	0.86
16p12.1_dup	renal_failure	373722	190	7922	2	4.0	0.50	0.42	0.62	0.13	1.78	1532	0.87
Potocki_Lupski	renal_failure	373537	5	7920	0	0.1	0.00	0.44	3.94	0.03	37.28	1584	0.87
8p23.1_dup	renal_failure	373537	5	7920	0	0.1	0.00	0.46	3.63	0.03	33.08	1647	0.88
13q12del_CRYL1	renal_failure	373887	355	7928	8	7.5	1.06	0.51	1.27	0.59	2.36	1782	0.89
TAR_dup	renal_failure	373930	398	7930	10	8.4	1.18	0.51	1.24	0.62	2.17	1800	0.89
15q11q13dup_BP3_BP5	renal_failure	373541	9	7920	0	0.2	0.00	0.54	2.83	0.02	23.65	1854	0.91
10q23_dup	renal_failure	373539	7	7920	0	0.1	0.00	0.55	2.79	0.02	26.07	1878	0.91
13q12dup_CRYL1	renal_failure	373542	10	7920	0	0.2	0.00	0.57	2.55	0.02	20.69	1946	0.92
2q21.1_del	renal_failure	373571	39	7921	1	0.8	1.20	0.60	1.60	0.18	6.07	2015	0.94
15q11q13del_BP3-BP4	renal_failure	373546	14	7920	0	0.3	0.00	0.62	2.21	0.02	16.69	2077	0.94
1q21.1_dup	renal_failure	373696	164	7924	4	3.5	1.15	0.64	1.27	0.42	2.89	2114	0.94
PWS_dup	renal_failure	373548	16	7920	0	0.3	0.00	0.66	2.04	0.02	16.10	2173	0.95
2q13_del	renal_failure	373582	50	7921	1	1.1	0.94	0.67	1.46	0.16	5.50	2195	0.95
15q11q13dup_BP3_BP4	renal_failure	373581	49	7921	1	1.0	0.96	0.67	1.46	0.16	5.48	2211	0.95
15q13.3del(CHRNA7)	renal_failure	373542	10	7920	0	0.2	0.00	0.68	1.95	0.01	15.94	2230	0.95
17q11.2_del_NF1	renal_failure	373541	9	7920	0	0.2	0.00	0.68	1.94	0.01	15.94	2234	0.95
15q11.2_dup	renal_failure	375412	1880	7957	37	39.8	0.93	0.68	0.93	0.66	1.27	2241	0.95
WBS_dup	renal_failure	373545	13	7920	0	0.3	0.00	0.76	1.61	0.01	12.64	2460	0.97
22q11.2distal_dup	renal_failure	373544	12	7920	0	0.3	0.00	0.82	1.42	0.01	11.13	2635	0.97
2q11.2_del	renal_failure	373561	29	7920	0	0.6	0.00	0.86	0.78	0.01	5.66	2756	0.97
15q13.3dup(CHRNA7)	renal_failure	376316	2784	7979	59	59.0	1.00	0.87	1.02	0.78	1.31	2790	0.98
10q11.21q11.23_del	renal_failure	373587	55	7921	1	1.2	0.86	0.89	1.13	0.13	4.23	2829	0.98
2q11.2_dup	renal_failure	373558	26	7920	0	0.6	0.00	0.98	1.04	0.01	7.55	3084	0.99
16p11.2distal_dup	renal_failure	373657	125	7922	2	2.7	0.76	1.00	1.00	0.21	2.88	3129	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p12.1_del	respiratory	360911	209	20776	26	12.0	2.03	0.00069	2.18	1.42	3.21	37	0.059*
16p11.2_del	respiratory	360790	88	20765	15	5.1	2.68	0.0023	2.59	1.44	4.37	60	0.12
TAR_del	respiratory	360763	61	20758	8	3.5	2.13	0.024	2.54	1.14	4.98	212	0.36
17q12_dup	respiratory	360790	88	20761	11	5.1	2.04	0.034	2.08	1.06	3.72	260	0.41
1q21.1_dup	respiratory	360854	152	20766	16	8.7	1.75	0.037	1.80	1.04	2.92	274	0.43
13q12del_CRYL1	respiratory	361038	336	20777	27	19.3	1.37	0.040	1.55	1.02	2.24	287	0.43
2q13dup(NPHP1)	respiratory	362450	1748	20870	120	100.7	1.18	0.063	1.20	0.99	1.44	371	0.53
22q11.2_dup	respiratory	360945	243	20773	23	14.0	1.59	0.076	1.52	0.95	2.29	434	0.55
13q12.12_del	respiratory	360778	76	20758	8	4.4	1.75	0.12	1.86	0.84	3.59	579	0.63
8p23.1_dup	respiratory	360706	4	20751	1	0.2	3.68	0.13	5.47	0.55	29.83	620	0.64
10q11.21q11.23_dup	respiratory	360738	36	20754	4	2.1	1.84	0.13	2.34	0.75	5.69	634	0.64
16p13.11_del	respiratory	360816	114	20760	10	6.6	1.48	0.15	1.66	0.82	2.99	679	0.67
15q24_dup	respiratory	360709	7	20751	1	0.4	2.30	0.15	4.55	0.48	21.08	700	0.69
2q21.1_del	respiratory	360738	36	20754	4	2.1	1.84	0.21	2.01	0.64	4.87	863	0.74
13q12.12_dup	respiratory	360899	197	20765	15	11.3	1.30	0.24	1.38	0.79	2.25	978	0.78
17p12_HNPP_del	respiratory	360913	211	20758	8	12.1	0.67	0.25	0.68	0.32	1.27	993	0.78
17q12_del	respiratory	360708	6	20751	1	0.3	2.63	0.26	3.19	0.33	15.74	1037	0.79
15q11.2_dup	respiratory	362503	1801	20866	116	103.7	1.11	0.27	1.11	0.92	1.34	1054	0.80
7q11.23_dup_distal	respiratory	360723	21	20752	2	1.2	1.60	0.29	2.21	0.44	6.96	1118	0.80
1q21.1_del	respiratory	360805	103	20753	3	5.9	0.52	0.29	0.59	0.16	1.48	1130	0.81
13q12dup_CRYL1	respiratory	360711	9	20751	1	0.5	1.84	0.30	2.82	0.30	12.36	1154	0.81
17q11.2_del_NF1	respiratory	360710	8	20751	1	0.5	2.04	0.33	2.67	0.28	11.99	1242	0.82
16p12.1_dup	respiratory	360887	185	20757	7	10.6	0.67	0.33	0.71	0.31	1.38	1269	0.82
15q11q13dup_BP3_BP4	respiratory	360748	46	20754	4	2.6	1.47	0.34	1.65	0.54	3.94	1303	0.83
2q13del(NPHP1)	respiratory	362916	2214	20866	116	127.3	0.92	0.36	0.92	0.76	1.10	1352	0.84
2q11.2_dup	respiratory	360726	24	20752	2	1.4	1.41	0.38	1.91	0.38	5.91	1388	0.85
2q13_del	respiratory	360749	47	20754	4	2.7	1.44	0.38	1.60	0.52	3.81	1396	0.85
15q11q13del_BP3-BP4	respiratory	360715	13	20751	1	0.7	1.31	0.41	2.21	0.24	9.13	1485	0.86
2q13_dup	respiratory	360771	69	20752	2	4.0	0.52	0.42	0.62	0.13	1.79	1520	0.87
15q13.3_dup	respiratory	360910	208	20766	16	12.0	1.31	0.45	1.22	0.71	1.96	1610	0.87
17p12_CMT1A_dup	respiratory	360810	108	20758	8	6.2	1.27	0.45	1.33	0.61	2.52	1612	0.87
16p13.11_dup	respiratory	361438	736	20797	47	42.3	1.10	0.52	1.10	0.81	1.46	1825	0.90
15q13.3_del	respiratory	360736	34	20753	3	2.0	1.49	0.58	1.39	0.37	3.70	1956	0.93
2q11.2_del	respiratory	360729	27	20752	2	1.6	1.27	0.62	1.42	0.29	4.37	2058	0.94
22q11.2distal_dup	respiratory	360714	12	20750	0	0.7	0.00	0.66	0.56	0.00	4.29	2168	0.95
3q29_dup	respiratory	360707	5	20750	0	0.29	0.00	0.67	2.00	0.02	17.96	2212	0.95
WBS_dup	respiratory	360715	13	20750	0	0.7	0.00	0.70	0.60	0.00	4.57	2296	0.96
NRXN1_del	respiratory	360852	150	20757	7	8.6	0.82	0.72	0.87	0.38	1.70	2331	0.96
PWS_dup	respiratory	360718	16	20750	0	0.9	0.00	0.75	0.65	0.01	4.86	2420	0.97
15q13.3del(CHRNA7)	respiratory	360712	10	20750	0	0.6	0.00	0.77	0.67	0.01	5.37	2502	0.97
16p11.2_dup	respiratory	360825	123	20758	8	7.1	1.12	0.78	1.10	0.51	2.09	2540	0.97
16p11.2distal_dup	respiratory	360822	120	20757	7	6.9	1.01	0.80	1.10	0.48	2.16	2599	0.97
Potocki_Lupski	respiratory	360707	5	20750	0	0.3	0.00	0.81	1.45	0.01	13.11	2619	0.97
22q11.2distal_del	respiratory	360707	5	20750	0	0.3	0.00	0.82	1.43	0.01	13.21	2637	0.97
22q11.2_del	respiratory	360712	10	20750	0	0.6	0.00	0.82	0.73	0.01	5.84	2641	0.97
15q11.2_del	respiratory	362187	1485	20835	85	85.4	1.00	0.85	0.98	0.78	1.21	2726	0.97
16p11.2distal_del	respiratory	360753	51	20753	3	2.9	1.02	0.85	1.11	0.30	2.86	2752	0.97
2q21.1_dup	respiratory	360754	52	20753	3	3.0	1.00	0.88	1.09	0.30	2.81	2814	0.98
3q29_del	respiratory	360711	9	20750	0	0.5	0.00	0.89	0.82	0.01	6.52	2826	0.98
10q23_dup	respiratory	360709	7	20750	0	0.4	0.00	0.94	1.12	0.01	9.66	2982	0.99
15q11q13dup_BP3_BP5	respiratory	360711	9	20750	0	0.5	0.00	0.94	0.90	0.01	7.29	2993	0.99
10q11.21q11.23_del	respiratory	360755	53	20753	3	3.0	0.98	0.95	1.04	0.28	2.68	3009	0.99
TAR_dup	respiratory	361088	386	20772	22	22.2	0.99	0.99	1.00	0.63	1.50	3113	1.00
15q13.3dup(CHRNA7)	respiratory	363393	2691	20902	152	154.8	0.98	1.00	1.00	0.84	1.17	3131	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2_dup	sciatica	338444	103	43139	28	13.1	1.89	0.0010	2.12	1.37	3.17	39	0.083*
13q12.12_del	sciatica	338406	65	43130	19	8.3	2.00	0.0027	2.32	1.36	3.78	62	0.14
2q21.1_dup	sciatica	338395	54	43112	1	6.9	0.16	0.016	0.22	0.02	0.80	165	0.31
2q13_dup	sciatica	338410	69	43113	2	8.8	0.25	0.017	0.29	0.06	0.83	169	0.32
22q11.2_dup	sciatica	338588	247	43130	19	31.5	0.63	0.021	0.60	0.37	0.93	194	0.35
17p12_CMT1A_dup	sciatica	338451	110	43117	6	14.0	0.46	0.028	0.45	0.18	0.93	234	0.37
2q21.1_del	sciatica	338372	31	43120	9	4.0	1.99	0.040	2.28	1.04	4.55	293	0.43
TAR_dup	sciatica	338714	373	43146	35	47.5	0.76	0.096	0.75	0.52	1.05	511	0.59
16p13.11_del	sciatica	338445	104	43131	20	13.3	1.43	0.097	1.53	0.92	2.40	516	0.59
15q24_dup	sciatica	338347	6	43113	2	0.8	2.21	0.16	3.21	0.59	12.67	720	0.69
1q21.1_del	sciatica	338431	90	43127	16	11.5	1.34	0.23	1.40	0.80	2.31	918	0.77
15q11q13dup_BP3_BP4	sciatica	338383	42	43119	8	5.4	1.42	0.23	1.61	0.71	3.20	941	0.78
PWS_dup	sciatica	338357	16	43111	0	2.0	0.00	0.24	0.26	0.00	1.91	955	0.78
3q29_del	sciatica	338348	7	43113	2	0.9	1.97	0.24	2.54	0.47	9.55	977	0.78
16p11.2distal_dup	sciatica	338449	108	43130	19	13.8	1.32	0.28	1.33	0.78	2.12	1090	0.80
15q13.3dup(CHRNA7)	sciatica	340850	2509	43445	334	319.8	1.04	0.29	1.07	0.95	1.19	1116	0.80
15q13.3del(CHRNA7)	sciatica	338349	8	43113	2	1.0	1.77	0.29	2.29	0.43	8.34	1125	0.81
16p12.1_dup	sciatica	338515	174	43129	18	22.2	0.83	0.31	0.78	0.46	1.24	1209	0.81
2q11.2_dup	sciatica	338363	22	43115	4	2.8	1.36	0.35	1.67	0.53	4.22	1312	0.83
17q12_dup	sciatica	338426	85	43125	14	10.8	1.25	0.35	1.32	0.72	2.23	1317	0.83
TAR_del	sciatica	338405	64	43116	5	8.2	0.64	0.37	0.69	0.25	1.50	1372	0.85
16p12.1_del	sciatica	338545	204	43142	31	26.0	1.17	0.37	1.19	0.80	1.71	1374	0.85
13q12.12_dup	sciatica	338525	184	43139	28	23.4	1.17	0.39	1.19	0.79	1.74	1437	0.85
13q12dup_CRYL1	sciatica	338351	10	43111	0	1.3	0.00	0.42	0.37	0.00	2.87	1524	0.86
17q11.2_del_NF1	sciatica	338350	9	43111	0	1.1	0.00	0.44	0.38	0.00	3.02	1588	0.87
15q11q13dup_BP3_BP5	sciatica	338350	9	43111	0	1.1	0.00	0.45	0.39	0.00	3.09	1620	0.88
2q13_del	sciatica	338385	44	43118	7	5.6	1.21	0.50	1.32	0.56	2.70	1758	0.89
16p13.11_dup	sciatica	339041	700	43194	83	89.2	0.94	0.50	0.93	0.73	1.15	1759	0.89
10q23_dup	sciatica	338347	6	43112	1	0.8	1.26	0.51	1.90	0.20	9.17	1790	0.89
17p12_HNPP_del	sciatica	338538	197	43133	22	25.1	0.89	0.56	0.88	0.55	1.33	1907	0.91
7q11.23_dup_distal	sciatica	338361	20	43114	3	2.5	1.15	0.58	1.39	0.37	3.84	1968	0.93
10q11.21q11.23_del	sciatica	338392	51	43116	5	6.5	0.79	0.59	0.79	0.29	1.75	1990	0.93
17q12_del	sciatica	338347	6	43112	1	0.8	1.26	0.61	1.62	0.17	7.78	2047	0.94
10q11.21q11.23_dup	sciatica	338378	37	43114	3	4.7	0.66	0.65	0.78	0.21	2.06	2164	0.95
15q11.2_del	sciatica	339738	1397	43284	173	178.0	0.97	0.67	0.97	0.82	1.13	2208	0.95
1q21.1_dup	sciatica	338488	147	43132	21	18.7	1.11	0.68	1.10	0.68	1.69	2246	0.95
NRXN1_del	sciatica	338479	138	43130	19	17.6	1.07	0.69	1.10	0.66	1.73	2272	0.95
15q13.3_del	sciatica	338373	32	43116	5	4.1	1.20	0.70	1.20	0.43	2.74	2297	0.96
22q11.2distal_del	sciatica	338346	5	43111	0	0.6	0.00	0.73	0.62	0.00	5.55	2374	0.96
16p11.2_del	sciatica	338431	90	43124	13	11.5	1.12	0.74	1.10	0.59	1.89	2404	0.96
2q13dup(NPHP1)	sciatica	339993	1652	43327	216	210.5	1.02	0.76	1.02	0.88	1.18	2454	0.96
15q11.2_dup	sciatica	340035	1694	43334	223	215.9	1.03	0.76	1.02	0.89	1.17	2464	0.97
Potocki_Lupski	sciatica	338346	5	43111	0	0.6	0.00	0.79	0.69	0.01	6.09	2559	0.97
22q11.2_del	sciatica	338350	9	43112	1	1.1	0.88	0.81	1.24	0.13	5.42	2616	0.97
8p23.1_dup	sciatica	338346	5	43111	0	0.6	0.00	0.83	0.73	0.01	6.46	2655	0.97
13q12del_CRYL1	sciatica	338666	325	43149	38	41.4	0.93	0.85	0.97	0.68	1.34	2748	0.97
WBS_dup	sciatica	338353	12	43112	1	1.5	0.68	0.88	0.88	0.10	3.68	2820	0.98
3q29_dup	sciatica	338346	5	43111	0	0.64	0.00	0.89	0.82	0.01	7.46	2854	0.98
22q11.2distal_dup	sciatica	338352	11	43112	1	1.4	0.74	0.90	0.90	0.10	3.78	2872	0.98
15q13.3_dup	sciatica	338539	198	43137	26	25.2	1.03	0.91	1.02	0.67	1.51	2911	0.98
15q11q13del_BP3-BP4	sciatica	338354	13	43112	1	1.7	0.63	0.95	0.95	0.10	3.88	3016	0.99
2q11.2_del	sciatica	338367	26	43114	3	3.3	0.92	0.97	1.02	0.27	2.75	3062	0.99
16p11.2distal_del	sciatica	338389	48	43117	6	6.1	0.98	0.97	1.02	0.40	2.15	3064	0.99
2q13del(NPHP1)	sciatica	340407	2066	43375	264	263.3	1.00	0.97	1.00	0.88	1.14	3076	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
17p12_CMT1A_dup	stroke_derived2017	372741	107	8827	9	2.5	3.36	0.0013	3.70	1.76	6.92	43	0.094*
16p13.11_del	stroke_derived2017	372750	116	8826	8	2.7	2.79	0.0049	3.28	1.49	6.29	78	0.20
15q13.3_dup	stroke_derived2017	372846	212	8830	12	5.0	2.32	0.015	2.23	1.19	3.81	156	0.30
22q11.2distal_dup	stroke_derived2017	372644	10	8820	2	0.2	7.21	0.021	7.74	1.46	27.83	187	0.35
22q11.2distal_del	stroke_derived2017	372638	4	8819	1	0.1	8.65	0.028	15.56	1.46	94.10	233	0.37
7q11.23_dup_distal	stroke_derived2017	372655	21	8820	2	0.5	3.76	0.039	5.65	1.12	18.14	282	0.43
10q11.21q11.23_del	stroke_derived2017	372686	52	8822	4	1.2	3.09	0.040	3.31	1.07	7.96	291	0.43
1q21.1_dup	stroke_derived2017	372794	160	8826	8	3.8	2.06	0.052	2.16	0.99	4.09	335	0.48
16p11.2distal_dup	stroke_derived2017	372755	121	8824	6	2.9	2.04	0.054	2.42	0.98	4.98	350	0.49
15q11q13dup_BP3_BP4	stroke_derived2017	372681	47	8821	3	1.1	2.60	0.073	3.23	0.88	8.48	417	0.55
3q29_del	stroke_derived2017	372642	8	8819	1	0.2	4.81	0.083	6.91	0.72	32.18	465	0.56
15q11q13del_BP3-BP4	stroke_derived2017	372647	13	8819	1	0.3	3.09	0.089	6.36	0.69	26.38	484	0.58
PWS_dup	stroke_derived2017	372649	15	8819	1	0.4	2.70	0.094	6.20	0.66	26.40	506	0.58
16p11.2distal_del	stroke_derived2017	372685	51	8821	3	1.2	2.40	0.10	2.86	0.78	7.41	532	0.60
15q13.3del(CHRNA7)	stroke_derived2017	372643	9	8819	1	0.2	4.33	0.11	5.82	0.61	26.91	547	0.62
16p11.2_dup	stroke_derived2017	372759	125	8824	6	3.0	1.98	0.13	1.98	0.80	4.07	626	0.63
WBS_dup	stroke_derived2017	372646	12	8819	1	0.3	3.33	0.14	4.88	0.52	21.11	655	0.65
NRXN1_del	stroke_derived2017	372785	151	8824	6	3.6	1.65	0.16	1.88	0.76	3.83	707	0.69
2q13_dup	stroke_derived2017	372705	71	8818	0	1.7	0.00	0.28	0.29	0.00	2.05	1112	0.80
3q29_dup	stroke_derived2017	372639	5	8818	0	0.12	0.00	0.33	6.28	0.05	57.40	1247	0.82
16p11.2_del	stroke_derived2017	372733	99	8822	4	2.3	1.68	0.37	1.61	0.52	3.76	1357	0.84
2q13dup(NPHP1)	stroke_derived2017	374465	1831	8855	37	43.3	0.86	0.39	0.87	0.62	1.18	1423	0.85
15q11.2_dup	stroke_derived2017	374501	1867	8868	50	44.2	1.13	0.39	1.14	0.85	1.49	1427	0.85
13q12.12_del	stroke_derived2017	372715	81	8821	3	1.9	1.54	0.39	1.67	0.46	4.26	1431	0.85
15q24_dup	stroke_derived2017	372642	8	8818	0	0.2	0.00	0.42	4.11	0.03	33.81	1535	0.87
2q13del(NPHP1)	stroke_derived2017	374905	2271	8877	59	53.8	1.10	0.43	1.11	0.85	1.43	1559	0.87
22q11.2_dup	stroke_derived2017	372892	258	8826	8	6.1	1.30	0.44	1.33	0.61	2.49	1574	0.87
2q13_del	stroke_derived2017	372685	51	8818	0	1.2	0.00	0.48	0.42	0.00	2.94	1689	0.88
8p23.1_dup	stroke_derived2017	372639	5	8818	0	0.1	0.00	0.48	3.41	0.03	30.85	1709	0.88
Potocki Lupski	stroke_derived2017	372639	5	8818	0	0.1	0.00	0.48	3.43	0.03	32.26	1711	0.88
1q21.1_del	stroke_derived2017	372739	105	8819	1	2.5	0.41	0.54	0.62	0.07	2.27	1850	0.91
10q23_dup	stroke_derived2017	372641	7	8818	0	0.2	0.00	0.55	2.73	0.02	24.87	1895	0.92
17q12_dup	stroke_derived2017	372730	96	8821	3	2.3	1.31	0.56	1.40	0.39	3.54	1914	0.91
15q11q13dup_BP3_BP5	stroke_derived2017	372643	9	8818	0	0.2	0.00	0.60	2.39	0.02	19.87	2009	0.93
13q12dup_CRYL1	stroke_derived2017	372644	10	8818	0	0.2	0.00	0.63	2.20	0.02	17.56	2093	0.94
15q11.2_del	stroke_derived2017	374170	1536	8852	34	36.3	0.94	0.63	0.92	0.64	1.27	2098	0.94
17q12_del	stroke_derived2017	372641	7	8818	0	0.2	0.00	0.63	2.19	0.02	19.06	2107	0.94
16p12.1_dup	stroke_derived2017	372823	189	8821	3	4.5	0.68	0.65	0.79	0.22	1.96	2142	0.95
2q21.1_del	stroke_derived2017	372673	39	8819	1	0.9	1.08	0.67	1.45	0.16	5.52	2218	0.95
22q11.2_del	stroke_derived2017	372644	10	8818	0	0.2	0.00	0.68	1.93	0.01	16.08	2250	0.95
16p12.1_del	stroke_derived2017	372863	229	8824	6	5.4	1.10	0.69	1.17	0.48	2.37	2279	0.95
17q11.2_del_NF1	stroke_derived2017	372643	9	8818	0	0.2	0.00	0.70	1.86	0.01	14.93	2286	0.96
10q11.21q11.23_dup	stroke_derived2017	372674	40	8818	0	0.9	0.00	0.72	0.62	0.00	4.45	2341	0.96
17p12_HNPP_del	stroke_derived2017	372849	215	8822	4	5.1	0.79	0.72	0.85	0.28	1.93	2370	0.96
13q12del_CRYL1	stroke_derived2017	372991	357	8824	6	8.4	0.72	0.73	0.87	0.36	1.75	2376	0.96
TAR_dup	stroke_derived2017	373032	398	8828	10	9.4	1.06	0.73	1.11	0.56	1.96	2392	0.96
13q12.12_dup	stroke_derived2017	372841	207	8823	5	4.9	1.02	0.74	1.16	0.43	2.46	2401	0.96
2q11.2_del	stroke_derived2017	372663	29	8818	0	0.7	0.00	0.76	0.67	0.01	4.83	2467	0.97
15q13.3_del	stroke_derived2017	372670	36	8819	1	0.9	1.17	0.78	1.28	0.14	4.92	2514	0.97
16p13.11_dup	stroke_derived2017	373398	764	8837	19	18.1	1.05	0.79	1.07	0.65	1.63	2543	0.97
2q21.1_dup	stroke_derived2017	372688	54	8819	1	1.3	0.79	0.91	1.09	0.12	4.08	2912	0.98
2q11.2_dup	stroke_derived2017	372660	26	8818	0	0.6	0.00	0.95	0.92	0.01	6.63	3011	0.99
TAR_del	stroke_derived2017	372702	68	8819	1	1.6	0.63	0.96	0.96	0.11	3.56	3045	0.99
15q13.3dup(CHRNA7)	stroke_derived2017	375413	2779	8882	64	65.7	0.97	1.00	1.00	0.77	1.27	3132	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
22q11.2distal_del	thyroid	354315	3	27142	2	0.2	5.62	0.016	11.34	1.68	68.80	162	0.30
TAR_del	thyroid	354371	59	27150	10	4.5	2.04	0.016	2.49	1.20	4.70	166	0.31
Potocki_Lupski	thyroid	354315	3	27142	2	0.2	5.62	0.022	9.47	1.44	55.75	201	0.34
2q13dup(NPHP1)	thyroid	356029	1717	27291	151	131.6	1.14	0.067	1.18	0.99	1.39	384	0.55
17q11.2_del_NF1	thyroid	354319	7	27142	2	0.5	3.12	0.071	4.82	0.85	19.76	411	0.54
15q24_dup	thyroid	354318	6	27142	2	0.5	3.51	0.077	4.67	0.82	20.08	439	0.55
15q11q13dup_BP3_BP5	thyroid	354319	7	27142	2	0.5	3.12	0.079	4.66	0.81	19.89	448	0.55
13q12.12_dup	thyroid	354503	191	27161	21	14.6	1.39	0.096	1.50	0.93	2.32	512	0.59
16p13.11_del	thyroid	354422	110	27154	14	8.4	1.59	0.12	1.60	0.88	2.71	594	0.63
22q11.2_dup	thyroid	354567	255	27151	11	19.5	0.58	0.12	0.64	0.33	1.11	599	0.63
NRXN1_del	thyroid	354454	142	27155	15	10.9	1.34	0.13	1.55	0.87	2.57	644	0.64
8p23.1_dup	thyroid	354316	4	27141	1	0.3	2.81	0.16	5.05	0.46	32.24	715	0.69
15q13.3dup(CHRNA7)	thyroid	356935	2623	27360	220	201.1	1.09	0.17	1.10	0.96	1.27	748	0.71
16p12.1_del	thyroid	354525	213	27162	22	16.3	1.32	0.17	1.38	0.86	2.10	758	0.72
17q12_dup	thyroid	354403	91	27148	8	7.0	1.14	0.24	1.58	0.72	3.07	953	0.78
22q11.2_del	thyroid	354321	9	27141	1	0.7	1.41	0.26	3.23	0.34	14.97	1016	0.79
17p12_HNPP_del	thyroid	354512	200	27159	19	15.3	1.22	0.32	1.28	0.77	2.01	1238	0.82
2q21.1_del	thyroid	354348	36	27144	4	2.8	1.41	0.33	1.71	0.54	4.26	1243	0.82
3q29_del	thyroid	354320	8	27141	1	0.6	1.56	0.34	2.67	0.27	13.06	1273	0.83
13q12.12_del	thyroid	354388	76	27148	8	5.8	1.34	0.34	1.44	0.65	2.82	1296	0.83
2q13_dup	thyroid	354377	65	27146	6	5.0	1.19	0.37	1.48	0.59	3.14	1384	0.85
2q13_del	thyroid	354359	47	27144	4	3.6	1.10	0.41	1.56	0.50	3.79	1490	0.86
16p13.11_dup	thyroid	355035	723	27200	60	55.4	1.08	0.44	1.11	0.84	1.44	1571	0.87
16p11.2_del	thyroid	354411	99	27144	4	7.6	0.55	0.47	0.71	0.23	1.66	1662	0.88
15q13.3_del	thyroid	354348	36	27141	1	2.8	0.38	0.49	0.59	0.07	2.25	1736	0.89
PWS_dup	thyroid	354328	16	27140	0	1.2	0.00	0.51	0.43	0.00	3.31	1777	0.89
22q11.2distal_dup	thyroid	354324	12	27140	0	0.9	0.00	0.51	0.43	0.00	3.46	1786	0.90
7q11.23_dup_distal	thyroid	354333	21	27142	2	1.6	1.22	0.56	1.51	0.30	4.90	1930	0.91
13q12dup_CRYL1	thyroid	354321	9	27141	1	0.7	1.41	0.58	1.70	0.18	7.73	1948	0.93
10q11.21q11.23_del	thyroid	354365	53	27143	3	4.1	0.75	0.61	0.76	0.21	1.99	2051	0.94
WBS_dup	thyroid	354325	13	27140	0	1.0	0.00	0.61	0.52	0.00	4.11	2054	0.94
17p12_CMT1A_dup	thyroid	354419	107	27149	9	8.2	1.09	0.63	1.19	0.56	2.22	2088	0.94
TAR_dup	thyroid	354691	379	27169	29	29.0	1.00	0.63	1.10	0.74	1.58	2090	0.94
15q11q13del_BP3-BP4	thyroid	354325	13	27141	1	1.0	1.00	0.63	1.57	0.17	6.88	2092	0.94
1q21.1_del	thyroid	354412	100	27146	6	7.7	0.80	0.68	0.85	0.34	1.76	2235	0.95
13q12del_CRYL1	thyroid	354649	337	27166	26	25.8	1.01	0.68	1.09	0.71	1.60	2249	0.95
2q11.2_dup	thyroid	354336	24	27142	2	1.8	1.08	0.69	1.32	0.26	4.24	2275	0.95
15q11.2_del	thyroid	355772	1460	27250	110	111.8	0.98	0.70	1.04	0.85	1.26	2301	0.96
3q29_dup	thyroid	354317	5	27140	0	0.38	0.00	0.73	1.79	0.01	22.57	2380	0.96
15q11q13dup_BP3_BP4	thyroid	354359	47	27143	3	3.6	0.84	0.77	1.19	0.32	3.18	2485	0.97
17q12_del	thyroid	354319	7	27140	0	0.5	0.00	0.83	1.41	0.01	12.20	2658	0.97
10q23_dup	thyroid	354319	7	27140	0	0.5	0.00	0.84	1.37	0.01	12.62	2699	0.97
15q13.3_dup	thyroid	354524	212	27152	12	16.2	0.75	0.85	0.95	0.50	1.62	2747	0.97
2q11.2_del	thyroid	354340	28	27141	1	2.1	0.48	0.86	0.86	0.09	3.38	2757	0.97
16p12.1_dup	thyroid	354491	179	27153	13	13.7	0.95	0.86	0.95	0.52	1.60	2763	0.97
1q21.1_dup	thyroid	354468	156	27152	12	11.9	1.00	0.87	1.05	0.56	1.81	2783	0.98
10q11.21q11.23_dup	thyroid	354350	38	27142	2	2.9	0.70	0.89	1.10	0.22	3.39	2842	0.98
15q11.2_dup	thyroid	356091	1779	27278	138	136.3	1.01	0.90	1.01	0.84	1.20	2875	0.98
16p11.2distal_del	thyroid	354363	51	27143	3	3.9	0.78	0.90	1.07	0.29	2.81	2879	0.98
16p11.2_dup	thyroid	354435	123	27148	8	9.4	0.86	0.91	0.96	0.44	1.83	2893	0.98
2q13del(NPHP1)	thyroid	356474	2162	27308	168	165.6	1.01	0.91	1.01	0.86	1.18	2908	0.98
15q13.3del(CHRNA7)	thyroid	354322	10	27140	0	0.8	0.00	0.94	0.89	0.01	7.61	2970	0.99
16p11.2distal_dup	thyroid	354431	119	27148	8	9.1	0.89	0.94	0.97	0.44	1.86	2984	0.99
2q21.1_dup	thyroid	354364	52	27143	3	4.0	0.77	0.95	1.03	0.28	2.72	3019	0.99

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p12.1_del	ureter_bladder	333111	186	48576	49	27.1	1.64	0.00033	1.85	1.34	2.52	29	0.036*
2q13_del	ureter_bladder	332962	37	48541	14	5.4	2.16	0.0018	2.91	1.52	5.27	53	0.11
16p13.11_dup	ureter_bladder	333580	655	48655	128	95.5	1.29	0.0023	1.36	1.12	1.64	59	0.12
17p12_CMT1A_dup	ureter_bladder	333016	91	48552	25	13.3	1.69	0.0054	1.96	1.23	3.01	81	0.21
15q24_dup	ureter_bladder	332930	5	48530	3	0.7	2.95	0.020	6.00	1.37	22.99	182	0.35
16p11.2_dup	ureter_bladder	333030	105	48553	26	15.3	1.56	0.022	1.70	1.08	2.58	197	0.35
16p11.2_del	ureter_bladder	333008	83	48547	20	12.1	1.53	0.034	1.76	1.05	2.81	257	0.41
17q12_del	ureter_bladder	332929	4	48530	3	0.6	3.37	0.038	5.08	1.11	21.55	277	0.43
TAR_del	ureter_bladder	332980	55	48541	14	8.0	1.59	0.043	1.90	1.02	3.33	302	0.45
15q11q13del_BP3-BP4	ureter_bladder	332935	10	48531	4	1.5	2.25	0.050	3.40	1.00	9.82	330	0.47
15q11.2_dup	ureter_bladder	334571	1646	48798	271	240.1	1.11	0.053	1.14	1.00	1.29	344	0.48
16p13.11_del	ureter_bladder	333027	102	48549	22	14.9	1.39	0.068	1.57	0.97	2.44	391	0.55
16p11.2distal_del	ureter_bladder	332968	43	48538	11	6.3	1.60	0.077	1.88	0.93	3.50	438	0.55
Potocki_Lupski	ureter_bladder	332928	3	48529	2	0.4	3.14	0.085	4.77	0.78	24.94	470	0.57
15q13.3del(CHRNA7)	ureter_bladder	332932	7	48530	3	1.0	2.36	0.097	3.28	0.78	11.27	517	0.59
10q23_dup	ureter_bladder	332930	5	48529	2	0.7	2.25	0.14	3.55	0.62	15.48	668	0.66
16p11.2distal_dup	ureter_bladder	333042	117	48537	10	17.1	0.62	0.16	0.65	0.32	1.17	723	0.69
10q11.21q11.23_dup	ureter_bladder	332958	33	48534	7	4.8	1.38	0.18	1.78	0.74	3.76	790	0.73
PWS_dup	ureter_bladder	332938	13	48530	3	1.9	1.47	0.19	2.37	0.60	7.12	819	0.74
17q12_dup	ureter_bladder	333007	82	48544	17	12.0	1.35	0.20	1.42	0.82	2.34	852	0.74
17p12_HNPP_del	ureter_bladder	332122	197	48549	22	28.7	0.79	0.20	0.76	0.48	1.15	860	0.75
1q21.1_del	ureter_bladder	333014	89	48544	17	13.0	1.26	0.21	1.41	0.82	2.31	870	0.75
2q11.2_dup	ureter_bladder	332946	21	48532	5	3.1	1.51	0.22	1.88	0.66	4.54	900	0.76
WBS_dup	ureter_bladder	332938	13	48527	0	1.9	0.00	0.23	0.25	0.00	1.92	937	0.78
1q21.1_dup	ureter_bladder	333067	142	48553	26	20.7	1.22	0.25	1.29	0.83	1.92	1010	0.78
22q11.2_del	ureter_bladder	332933	8	48529	2	1.2	1.57	0.28	2.35	0.44	8.78	1097	0.80
16p12.1_dup	ureter_bladder	333098	173	48546	19	25.2	0.78	0.30	0.78	0.47	1.22	1143	0.81
15q11.2_del	ureter_bladder	334307	1382	48715	188	201.4	0.94	0.34	0.93	0.79	1.08	1271	0.83
13q12.12_del	ureter_bladder	333001	76	48535	8	11.1	0.75	0.36	0.72	0.33	1.40	1330	0.84
22q11.2distal_del	ureter_bladder	332929	4	48528	1	0.6	1.57	0.36	2.60	0.26	14.57	1337	0.84
22q11.2_dup	ureter_bladder	333154	229	48564	37	33.4	1.09	0.43	1.15	0.80	1.61	1555	0.87
15q11q13dup_BP3_BP5	ureter_bladder	332934	9	48527	0	1.3	0.00	0.47	0.40	0.00	3.20	1655	0.88
13q12.12_dup	ureter_bladder	333108	183	48556	29	26.7	1.08	0.53	1.13	0.75	1.65	1848	0.90
15q11q13dup_BP3_BP4	ureter_bladder	332968	43	48534	7	6.3	1.10	0.61	1.23	0.52	2.54	2031	0.94
2q13dup(NPHP1)	ureter_bladder	334563	1638	48757	230	238.7	0.97	0.62	0.97	0.84	1.11	2063	0.94
22q11.2distal_dup	ureter_bladder	332935	10	48529	2	1.5	1.31	0.64	1.41	0.27	4.92	2119	0.94
8p23.1_dup	ureter_bladder	332930	5	48527	0	0.7	0.00	0.66	0.55	0.00	4.92	2186	0.95
10q11.21q11.23_del	ureter_bladder	332973	48	48535	8	7.0	1.12	0.69	1.16	0.52	2.30	2273	0.95
15q13.3_del	ureter_bladder	332958	33	48531	4	4.8	0.85	0.73	0.84	0.27	2.06	2378	0.96
TAR_dup	ureter_bladder	333280	355	48580	53	51.7	1.02	0.76	1.05	0.77	1.38	2482	0.97
2q21.1_dup	ureter_bladder	332974	49	48533	6	7.1	0.86	0.77	0.89	0.35	1.88	2494	0.97
7q11.23_dup_distal	ureter_bladder	332946	21	48529	2	3.1	0.68	0.78	0.83	0.17	2.61	2525	0.97
15q13.3_dup	ureter_bladder	333119	194	48557	30	28.3	1.05	0.80	1.05	0.70	1.52	2600	0.97
NRXN1_del	ureter_bladder	333062	137	48547	20	20.0	1.00	0.82	1.06	0.64	1.65	2636	0.97
2q13_dup	ureter_bladder	332987	62	48536	9	9.0	1.00	0.85	1.07	0.50	2.03	2736	0.97
3q29_del	ureter_bladder	332933	8	48528	1	1.2	0.87	0.86	1.17	0.12	5.27	2767	0.98
2q13del(NPHP1)	ureter_bladder	334957	2032	48825	298	296.2	1.01	0.88	1.01	0.89	1.14	2808	0.98
2q11.2_del	ureter_bladder	332951	26	48530	3	3.8	0.81	0.89	0.92	0.25	2.50	2828	0.98
3q29_dup	ureter_bladder	332930	5	48527	0	0.73	0.00	0.89	0.82	0.01	7.37	2836	0.98
13q12del_CRYL1	ureter_bladder	333244	319	48571	44	46.5	0.95	0.90	1.02	0.74	1.39	2863	0.98
15q13.3dup(CHRNA7)	ureter_bladder	335408	2483	48887	360	361.9	1.00	0.90	1.01	0.90	1.12	2883	0.98
13q12dup_CRYL1	ureter_bladder	332934	9	48528	1	1.3	0.79	0.91	1.11	0.12	4.87	2897	0.98
2q21.1_del	ureter_bladder	332960	35	48532	5	5.1	0.98	0.95	1.03	0.37	2.34	3026	0.99
17q11.2_del_NF1	ureter_bladder	332933	8	48528	1	1.2	0.87	0.99	1.01	0.11	4.54	3108	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2distal_del	uterine_problem	339206	42	42300	12	5.2	2.00	0.0020	3.60	1.63	7.97	55	0.11
16p12.1_del	uterine_problem	339361	197	42326	38	24.6	1.46	0.016	1.61	1.10	2.33	164	0.31
13q12.12_del	uterine_problem	339233	69	42303	15	8.6	1.61	0.044	1.93	1.02	3.50	308	0.45
16p11.2distal_dup	uterine_problem	339283	119	42296	8	14.8	0.57	0.059	0.53	0.24	1.02	360	0.51
7q11.23_dup_distal	uterine_problem	339187	23	42288	0	2.9	0.00	0.060	0.14	0.00	1.06	364	0.52
3q29_dup	uterine_problem	339168	4	42289	1	0.50	1.80	0.078	1759.05	0.72	18361.09	443	0.55
2q21.1_dup	uterine_problem	339211	47	42296	8	5.9	1.31	0.10	2.07	0.85	4.68	535	0.60
17q11.2_del_NF1	uterine_problem	339171	7	42290	2	0.9	2.00	0.13	4.14	0.64	26.87	625	0.64
16p11.2_del	uterine_problem	339253	89	42302	14	11.1	1.23	0.14	1.62	0.84	2.95	672	0.66
16p12.1_dup	uterine_problem	339339	175	42305	17	21.8	0.80	0.18	0.72	0.42	1.16	788	0.73
2q21.1_del	uterine_problem	339198	34	42294	6	4.2	1.35	0.27	1.72	0.63	4.18	1050	0.80
15q11.2_del	uterine_problem	340559	1395	42463	175	173.9	1.01	0.30	1.09	0.92	1.29	1171	0.81
22q11.2distal_dup	uterine_problem	339176	12	42288	0	1.5	0.00	0.31	0.29	0.00	2.41	1179	0.81
15q24_dup	uterine_problem	339172	8	42288	0	1.0	0.00	0.31	0.29	0.00	2.44	1190	0.82
17p12_HNPP_del	uterine_problem	339364	200	42307	19	24.9	0.78	0.32	0.79	0.47	1.25	1240	0.82
15q13.3_dup	uterine_problem	339374	210	42302	14	26.2	0.56	0.33	0.76	0.42	1.30	1258	0.82
1q21.1_del	uterine_problem	339255	91	42303	15	11.3	1.28	0.35	1.32	0.72	2.31	1326	0.84
2q13dup(NPHP1)	uterine_problem	340841	1677	42479	191	209.0	0.92	0.36	0.93	0.79	1.09	1348	0.85
1q21.1_dup	uterine_problem	339310	146	42310	22	18.2	1.18	0.38	1.24	0.75	1.97	1414	0.85
2q11.2_del	uterine_problem	339192	28	42289	1	3.5	0.31	0.38	0.50	0.05	2.09	1416	0.85
2q13_dup	uterine_problem	339230	66	42293	5	8.2	0.64	0.39	0.68	0.25	1.57	1442	0.85
2q13del(NPHP1)	uterine_problem	341219	2055	42563	275	256.3	1.06	0.42	1.06	0.92	1.21	1531	0.87
Potocki_Lupski	uterine_problem	339168	4	42289	1	0.5	1.80	0.43	2.36	0.22	17.74	1543	0.87
16p13.11_del	uterine_problem	339275	111	42301	13	13.8	0.95	0.45	0.80	0.43	1.39	1603	0.87
22q11.2_del	uterine_problem	339174	10	42288	0	1.2	0.00	0.45	0.37	0.00	3.49	1616	0.87
TAR_dup	uterine_problem	339527	363	42333	45	45.3	0.99	0.45	1.14	0.81	1.57	1625	0.88
PWS_dup	uterine_problem	339177	13	42291	3	1.6	1.69	0.46	1.65	0.40	5.55	1631	0.88
15q13.3del(CHRNA7)	uterine_problem	339173	9	42289	1	1.1	0.90	0.48	2.15	0.20	16.23	1693	0.88
17q12_dup	uterine_problem	339257	93	42294	6	11.6	0.55	0.48	0.75	0.29	1.62	1707	0.88
15q11q13dup_BP3_BP4	uterine_problem	339209	45	42293	5	5.6	0.90	0.48	1.44	0.49	3.64	1716	0.88
22q11.2distal_del	uterine_problem	339168	4	42289	1	0.5	1.80	0.49	2.10	0.19	15.87	1730	0.89
10q11.21q11.23_dup	uterine_problem	339200	36	42292	4	4.5	0.90	0.57	1.38	0.42	3.80	1934	0.92
22q11.2_dup	uterine_problem	339401	237	42317	29	29.5	0.98	0.63	1.11	0.72	1.65	2083	0.94
15q11.2_dup	uterine_problem	340871	1707	42498	210	212.8	0.99	0.65	0.97	0.83	1.12	2143	0.95
3q29_del	uterine_problem	339172	8	42289	1	1.0	1.00	0.67	1.54	0.15	9.37	2206	0.95
16p11.2_dup	uterine_problem	339281	117	42302	14	14.6	0.96	0.68	1.13	0.61	1.98	2238	0.95
15q13.3_del	uterine_problem	339197	33	42292	4	4.1	0.98	0.72	1.22	0.37	3.31	2353	0.96
13q12del_CRYL1	uterine_problem	339485	321	42330	42	40.0	1.04	0.73	1.06	0.75	1.48	2381	0.96
16p13.11_dup	uterine_problem	339865	701	42370	82	87.4	0.94	0.73	0.96	0.75	1.21	2387	0.96
2q11.2_dup	uterine_problem	339187	23	42291	3	2.9	1.04	0.76	1.21	0.31	3.66	2458	0.96
15q13.3dup(CHRNA7)	uterine_problem	341687	2523	42608	320	314.6	1.02	0.78	1.02	0.90	1.15	2532	0.97
10q23_dup	uterine_problem	339171	7	42288	0	0.9	0.00	0.79	0.67	0.00	8.30	2560	0.97
17q12_del	uterine_problem	339171	7	42288	0	0.9	0.00	0.82	0.71	0.01	8.71	2633	0.97
13q12.12_dup	uterine_problem	339351	187	42313	25	23.3	1.06	0.82	1.05	0.66	1.61	2649	0.97
2q13_del	uterine_problem	339211	47	42292	4	5.9	0.71	0.83	0.89	0.28	2.30	2670	0.97
15q11q13dup_BP3_BP5	uterine_problem	339172	8	42289	1	1.0	1.00	0.84	1.21	0.12	6.57	2709	0.97
10q11.21q11.23_del	uterine_problem	339214	50	42294	6	6.2	0.97	0.85	0.92	0.36	2.04	2731	0.97
TAR_del	uterine_problem	339226	62	42295	7	7.7	0.92	0.87	0.94	0.39	1.97	2784	0.98
15q11q13del_BP3-BP4	uterine_problem	339177	13	42289	1	1.6	0.64	0.88	0.87	0.09	4.16	2815	0.98
8p23.1_dup	uterine_problem	339169	5	42288	0	0.6	0.00	0.89	0.81	0.01	10.03	2851	0.98
NRXN1_del	uterine_problem	339305	141	42304	16	17.6	0.92	0.91	0.97	0.55	1.63	2899	0.98
13q12dup_CRYL1	uterine_problem	339173	9	42289	1	1.1	0.90	0.93	0.92	0.10	4.37	2945	0.98
17p12_CMT1A_dup	uterine_problem	339268	104	42300	12	13.0	0.93	0.94	0.98	0.50	1.76	2976	0.99
WBS_dup	uterine_problem	339176	12	42289	1	1.5	0.69	0.96	0.96	0.10	4.80	3043	0.99



CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
2q13del(NPHP1)	varicose_veins	371831	2231	11951	99	71.7	1.37	0.0027	1.38	1.12	1.68	63	0.14
2q13_dup	varicose_veins	369646	46	11857	5	1.5	3.16	0.010	3.98	1.45	8.88	125	0.26
10q11.21q11.23_dup	varicose_veins	369636	36	11856	4	1.2	3.22	0.013	4.52	1.45	10.92	140	0.28
15q11q13del_BP3-BP4	varicose_veins	369612	12	11854	2	0.4	4.60	0.025	6.93	1.35	23.40	214	0.36
3q29_dup	varicose_veins	369604	4	11853	1	0.13	6.44	0.026	15.47	1.53	85.75	221	0.36
17p12_HNPP_del	varicose_veins	369817	217	11854	2	7.0	0.29	0.049	0.35	0.07	1.00	325	0.47
17q12_del	varicose_veins	369606	6	11853	1	0.2	4.60	0.066	8.04	0.83	38.55	381	0.55
16p12.1_dup	varicose_veins	369782	182	11862	10	5.8	1.68	0.091	1.79	0.90	3.18	494	0.58
15q24_dup	varicose_veins	369607	7	11853	1	0.2	4.02	0.096	6.29	0.66	29.37	509	0.59
15q11.2_del	varicose_veins	371133	1533	11889	37	49.1	0.76	0.11	0.77	0.55	1.05	539	0.62
15q11q13dup_BP3_BP4	varicose_veins	369647	47	11855	3	1.5	1.93	0.15	2.48	0.68	6.42	688	0.68
22q11.2_dup	varicose_veins	369862	262	11856	4	8.4	0.48	0.16	0.55	0.18	1.23	726	0.69
WBS_dup	varicose_veins	369612	12	11853	1	0.4	2.48	0.20	3.74	0.40	15.77	839	0.74
16p11.2distal_del	varicose_veins	369651	51	11855	3	1.6	1.79	0.20	2.19	0.60	5.64	859	0.74
17p12_CMT1A_dup	varicose_veins	369711	111	11857	5	3.6	1.39	0.33	1.57	0.59	3.37	1268	0.82
TAR_del	varicose_veins	369666	66	11855	3	2.1	1.40	0.37	1.70	0.47	4.31	1380	0.85
1q21.1_dup	varicose_veins	369765	165	11855	3	5.3	0.57	0.39	0.65	0.18	1.61	1428	0.85
2q13_dup	varicose_veins	369668	68	11855	3	2.2	1.36	0.39	1.67	0.46	4.24	1429	0.85
13q12.12_del	varicose_veins	369683	83	11853	1	2.7	0.38	0.41	0.54	0.06	1.98	1502	0.86
7q11.23_dup_distal	varicose_veins	369622	22	11853	1	0.7	1.40	0.42	2.14	0.24	8.32	1509	0.87
15q13.3_del	varicose_veins	369637	37	11852	0	1.2	0.00	0.44	0.39	0.00	2.78	1591	0.87
13q12del_CRYL1	varicose_veins	369955	355	11860	8	11.4	0.71	0.48	0.79	0.37	1.46	1702	0.88
TAR_dup	varicose_veins	369994	394	11866	14	12.6	1.10	0.52	1.20	0.67	1.94	1805	0.90
8p23.1_dup	varicose_veins	369605	5	11852	0	0.2	0.00	0.52	3.01	0.02	26.72	1817	0.90
22q11.2distal_del	varicose_veins	369605	5	11852	0	0.2	0.00	0.55	2.73	0.02	24.64	1894	0.92
Potocki_Lupski	varicose_veins	369605	5	11852	0	0.2	0.00	0.57	2.60	0.02	22.99	1942	0.92
10q23_dup	varicose_veins	369607	7	11852	0	0.2	0.00	0.59	2.45	0.02	20.54	1985	0.93
1q21.1_del	varicose_veins	369704	104	11854	2	3.3	0.61	0.61	0.73	0.15	2.09	2028	0.94
15q13.3_dup	varicose_veins	369819	219	11857	5	7.0	0.72	0.65	0.82	0.31	1.74	2138	0.95
2q11.2_del	varicose_veins	369629	29	11852	0	0.9	0.00	0.65	0.55	0.00	3.93	2139	0.95
10q11.21q11.23_del	varicose_veins	369654	54	11854	2	1.7	1.15	0.66	1.35	0.28	3.94	2179	0.95
15q11.2_dup	varicose_veins	371461	1861	11908	56	59.7	0.94	0.69	0.95	0.72	1.22	2274	0.95
2q11.2_dup	varicose_veins	369626	26	11852	0	0.8	0.00	0.70	0.60	0.00	4.32	2292	0.96
2q13dup(NPHP1)	varicose_veins	371414	1814	11906	54	58.1	0.93	0.70	0.95	0.72	1.23	2302	0.96
15q13.3dup(CHRNA7)	varicose_veins	372353	2753	11942	90	88.3	1.02	0.72	1.04	0.84	1.27	2347	0.96
22q11.2_del	varicose_veins	369610	10	11852	0	0.3	0.00	0.74	1.69	0.01	13.24	2399	0.96
NRXN1_del	varicose_veins	369752	152	11857	5	4.9	1.02	0.74	1.16	0.43	2.46	2409	0.96
15q11q13dup_BP3_BP5	varicose_veins	369609	9	11852	0	0.3	0.00	0.74	1.67	0.01	13.38	2414	0.97
15q13.3del(CHRNA7)	varicose_veins	369610	10	11852	0	0.3	0.00	0.75	1.66	0.01	13.03	2417	0.97
16p11.2distal_dup	varicose_veins	369723	123	11856	4	3.9	1.01	0.75	1.17	0.39	2.67	2442	0.97
3q29_del	varicose_veins	369609	9	11852	0	0.3	0.00	0.75	1.64	0.01	13.02	2445	0.96
17q11.2_del_NF1	varicose_veins	369609	9	11852	0	0.3	0.00	0.78	1.55	0.01	12.31	2522	0.97
16p13.11_del	varicose_veins	369721	121	11855	3	3.9	0.78	0.79	0.87	0.24	2.17	2561	0.97
17q12_dup	varicose_veins	369697	97	11854	2	3.1	0.65	0.80	0.85	0.18	2.44	2582	0.97
16p11.2_del	varicose_veins	369700	100	11855	3	3.2	0.94	0.84	1.12	0.31	2.81	2692	0.97
2q21.1_del	varicose_veins	369639	39	11853	1	1.3	0.80	0.84	1.19	0.13	4.44	2712	0.97
13q12dup_CRYL1	varicose_veins	369610	10	11852	0	0.3	0.00	0.84	1.35	0.01	10.58	2715	0.97
16p11.2_dup	varicose_veins	369727	127	11856	4	4.1	0.98	0.86	1.09	0.36	2.49	2760	0.97
2q21.1_dup	varicose_veins	369654	54	11853	1	1.7	0.59	0.91	0.91	0.10	3.35	2896	0.98
13q12.12_dup	varicose_veins	369806	206	11858	6	6.6	0.91	0.92	0.96	0.39	1.94	2937	0.99
16p13.11_dup	varicose_veins	370359	759	11876	24	24.3	0.99	0.93	1.02	0.66	1.49	2949	0.98
22q11.2distal_dup	varicose_veins	369612	12	11852	0	0.4	0.00	0.94	1.12	0.01	8.59	2972	0.99
PWS_dup	varicose_veins	369616	16	11852	0	0.5	0.00	0.98	1.04	0.01	7.72	3082	0.99
16p12.1_del	varicose_veins	369828	228	11859	7	7.3	0.96	0.99	1.00	0.44	1.93	3111	1.00

CNV	Phenotype	N_con	N_CNV_con	N_case	N_CNV_case	Expected_N_CNVs	RR	pvalue	OR	ORL95	ORU95	rank	B-H FDR
16p11.2_dup	venous_thromboembolic_disease	367600	120	13983	11	4.6	2.29	0.0093	2.49	1.28	4.37	119	0.24
NRXN1_del	venous_thromboembolic_disease	367626	146	13983	11	5.6	1.91	0.027	2.13	1.10	3.72	227	0.37
TAR_dup	venous_thromboembolic_disease	367865	385	13995	23	14.6	1.54	0.031	1.64	1.05	2.43	249	0.39
15q11q13del_BP3-BP4	venous_thromboembolic_disease	367492	12	13974	2	0.5	3.90	0.036	5.94	1.16	20.00	270	0.41
16p12.1_del	venous_thromboembolic_disease	367701	221	13986	14	8.4	1.63	0.068	1.71	0.96	2.81	388	0.55
22q11.2_dup	venous_thromboembolic_disease	367731	251	13987	15	9.5	1.54	0.082	1.63	0.93	2.64	462	0.56
15q24_dup	venous_thromboembolic_disease	367487	7	13973	1	0.3	3.41	0.089	6.59	0.69	30.83	486	0.57
10q23_dup	venous_thromboembolic_disease	367486	6	13973	1	0.2	3.90	0.090	6.69	0.68	33.35	491	0.58
7q11.23_dup_distal	venous_thromboembolic_disease	367501	21	13974	2	0.8	2.37	0.14	3.18	0.64	9.97	660	0.65
3q29_del	venous_thromboembolic_disease	367488	8	13973	1	0.3	3.03	0.15	4.57	0.48	20.61	697	0.68
16p13.11_del	venous_thromboembolic_disease	367597	117	13979	7	4.4	1.54	0.20	1.69	0.73	3.31	836	0.74
TAR_del	venous_thromboembolic_disease	367545	65	13976	4	2.5	1.58	0.23	1.89	0.62	4.42	939	0.78
13q12.12_del	venous_thromboembolic_disease	367559	79	13977	5	3.0	1.63	0.24	1.76	0.65	3.82	963	0.78
2q13_del	venous_thromboembolic_disease	367528	48	13975	3	1.8	1.61	0.25	2.02	0.55	5.24	1004	0.78
17p12_HNPP_del	venous_thromboembolic_disease	367694	214	13977	5	8.1	0.62	0.30	0.66	0.25	1.40	1169	0.81
17p12_CMT1A_dup	venous_thromboembolic_disease	367590	110	13978	6	4.2	1.41	0.33	1.52	0.62	3.10	1263	0.82
2q13del(NPHP1)	venous_thromboembolic_disease	369716	2236	14066	94	85.1	1.10	0.39	1.10	0.89	1.34	1436	0.85
2q13dup(NPHP1)	venous_thromboembolic_disease	369273	1793	14047	75	68.2	1.10	0.39	1.11	0.87	1.39	1438	0.85
16p11.2_del	venous_thromboembolic_disease	367578	98	13977	5	3.7	1.33	0.39	1.49	0.55	3.22	1451	0.85
10q11.21q11.23_del	venous_thromboembolic_disease	367533	53	13975	3	2.0	1.46	0.44	1.58	0.43	4.07	1576	0.87
15q11.2_del	venous_thromboembolic_disease	368997	1517	14025	53	57.7	0.92	0.46	0.90	0.68	1.18	1628	0.88
15q13.3_dup	venous_thromboembolic_disease	367694	214	13982	10	8.1	1.22	0.47	1.27	0.64	2.25	1653	0.88
15q13.3_del	venous_thromboembolic_disease	367515	35	13974	2	1.3	1.48	0.47	1.67	0.34	5.03	1654	0.88
2q13_dup	venous_thromboembolic_disease	367550	70	13973	1	2.7	0.38	0.47	0.58	0.07	2.11	1656	0.88
3q29_dup	venous_thromboembolic_disease	367485	5	13972	0	0.19	0.00	0.49	3.38	0.03	30.89	1720	0.88
2q11.2_del	venous_thromboembolic_disease	367509	29	13972	0	1.1	0.00	0.54	0.46	0.00	3.30	1851	0.91
15q11q13dup_BP3_BP4	venous_thromboembolic_disease	367528	48	13974	2	1.8	1.09	0.59	1.45	0.30	4.29	1977	0.93
16p12.1_dup	venous_thromboembolic_disease	367664	184	13980	8	7.0	1.14	0.60	1.21	0.56	2.27	2005	0.93
16p11.2distal_dup	venous_thromboembolic_disease	367604	124	13975	3	4.7	0.64	0.60	0.76	0.21	1.91	2018	0.94
22q11.2distal_del	venous_thromboembolic_disease	367485	5	13972	0	0.2	0.00	0.61	2.35	0.02	21.79	2038	0.94
2q11.2_dup	venous_thromboembolic_disease	367506	26	13972	0	1.0	0.00	0.61	0.52	0.00	3.74	2044	0.94
15q11.2_dup	venous_thromboembolic_disease	369323	1843	14046	74	70.1	1.05	0.61	1.06	0.83	1.33	2049	0.94
Potocki_Lupski	venous_thromboembolic_disease	367485	5	13972	0	0.2	0.00	0.62	2.27	0.02	20.23	2061	0.94
8p23.1_dup	venous_thromboembolic_disease	367485	5	13972	0	0.2	0.00	0.63	2.20	0.02	19.56	2101	0.94
1q21.1_dup	venous_thromboembolic_disease	367641	161	13979	7	6.1	1.14	0.63	1.20	0.53	2.33	2104	0.94
15q13.3dup(CHRNA7)	venous_thromboembolic_disease	370225	2745	14070	98	104.3	0.94	0.67	0.96	0.78	1.16	2190	0.95
2q21.1_dup	venous_thromboembolic_disease	367534	54	13973	1	2.1	0.50	0.69	0.73	0.08	2.69	2264	0.95
16p11.2distal_del	venous_thromboembolic_disease	367532	52	13974	2	2.0	1.01	0.71	1.28	0.26	3.76	2315	0.96
17q12_del	venous_thromboembolic_disease	367487	7	13972	0	0.3	0.00	0.74	1.71	0.01	14.26	2394	0.96
16p13.11_dup	venous_thromboembolic_disease	368233	753	14002	30	28.6	1.05	0.75	1.06	0.72	1.50	2427	0.97
13q12del_CRYL1	venous_thromboembolic_disease	367832	352	13983	11	13.4	0.83	0.75	0.91	0.48	1.56	2432	0.97
15q11q13dup_BP3_BP5	venous_thromboembolic_disease	367489	9	13972	0	0.3	0.00	0.82	1.41	0.01	11.37	2651	0.97
22q11.2_del	venous_thromboembolic_disease	367490	10	13972	0	0.4	0.00	0.83	1.38	0.01	10.91	2680	0.97
1q21.1_del	venous_thromboembolic_disease	367583	103	13975	3	3.9	0.77	0.85	0.91	0.25	2.27	2746	0.97
10q11.21q11.23_dup	venous_thromboembolic_disease	367519	39	13973	1	1.5	0.68	0.86	1.16	0.13	4.37	2765	0.97
15q13.3del(CHRNA7)	venous_thromboembolic_disease	367490	10	13972	0	0.4	0.00	0.87	1.27	0.01	10.00	2800	0.98
13q12dup_CRYL1	venous_thromboembolic_disease	367490	10	13972	0	0.4	0.00	0.87	1.27	0.01	9.91	2801	0.98
17q11.2_del_NF1	venous_thromboembolic_disease	367489	9	13972	0	0.3	0.00	0.89	1.24	0.01	9.81	2833	0.98
22q11.2distal_dup	venous_thromboembolic_disease	367492	12	13972	0	0.5	0.00	0.92	0.86	0.01	6.60	2913	0.98
17q12_dup	venous_thromboembolic_disease	367576	96	13975	3	3.6	0.83	0.95	0.96	0.27	2.43	3004	0.99
13q12.12_dup	venous_thromboembolic_disease	367685	205	13979	7	7.8	0.90	0.95	0.98	0.43	1.89	3007	0.99
2q21.1_del	venous_thromboembolic_disease	367519	39	13973	1	1.5	0.68	0.95	0.95	0.11	3.57	3022	0.99
WBS_dup	venous_thromboembolic_disease	367493	13	13972	0	0.5	0.00	0.97	0.95	0.01	7.25	3068	0.99
PWS_dup	venous_thromboembolic_disease	367496	16	13972	0	0.6	0.00	0.99	0.98	0.01	7.33	3103	1.00

Sex	ICD10	Death_cause	CNV1	CNV2
FEMALE	C509	Cancer of the Breast	10q11.21q11.23del	
MALE	I269	Pulmonary embolism	10q11.21q11.23del	
FEMALE	A810	Creutzfeldt-Jakob disease	10q11.21q11.23dup	
MALE	I219	Myocardial Infarction	10q11.21q11.23dup	
MALE	R99	Unspecified	10q11.21q11.23dup	
FEMALE	C221	Cancer of the Liver	13q12.12del	
MALE	C349	Cancer of the lung	13q12.12del	15q11.2dup
FEMALE	C541	Cancer of the Endometrium	13q12.12del	15q13.3dup_CHRNA7
MALE	C61	Cancer of the Prostate	13q12.12del	
MALE	C911	Chronic lymphocytic leukaemia	13q12.12del	
MALE	E785	Hyperlipidaemia	13q12.12del	
MALE	W190	Unspecified fall	13q12.12del	
FEMALE	C189	Cancer of the Colon	13q12.12dup	
MALE	C259	Cancer of the Pancreas	13q12.12dup	
FEMALE	C259	Cancer of the Pancreas	13q12.12dup	
MALE	C349	Cancer of the lung	13q12.12dup	
MALE	C349	Cancer of the lung	13q12.12dup	
MALE	C349	Cancer of the lung	13q12.12dup	
FEMALE	C56	Cancer of the Ovary	13q12.12dup	
MALE	C64	Cancer of the Kidney	13q12.12dup	17p12dup_CMT1A
FEMALE	C719	Cancer of the Brain	13q12.12dup	
MALE	J841	Other interstitial pulmonary diseases	13q12.12dup	
MALE	X700	Suicide	13q12.12dup	
FEMALE	C509	Cancer of the Breast	13q12dup_CRYL1	
MALE	B182	Chronic viral hepatitis	15q11.2del	
MALE	C159	Cancer of the Oesophagus	15q11.2del	
MALE	C159	Cancer of the Oesophagus	15q11.2del	
MALE	C159	Cancer of the Oesophagus	15q11.2del	
FEMALE	C170	Cancer of the Small Intestine	15q11.2del	
MALE	C187	Cancer of the Colon	15q11.2del	
MALE	C189	Cancer of the Colon	15q11.2del	
FEMALE	C221	Cancer of the Liver	15q11.2del	
MALE	C240	Cancer of the Biliary Tract	15q11.2del	
MALE	C259	Cancer of the Pancreas	15q11.2del	
FEMALE	C260	Cancer of other digestive organs	15q11.2del	
MALE	C349	Cancer of the lung	15q11.2del	
FEMALE	C349	Cancer of the lung	15q11.2del	
MALE	C349	Cancer of the lung	15q11.2del	
MALE	C349	Cancer of the lung	15q11.2del	
FEMALE	C439	Cancer of the skin	15q11.2del	15q13.3dup_CHRNA7
MALE	C449	Cancer of the skin	15q11.2del	
FEMALE	C509	Cancer of the Breast	15q11.2del	
FEMALE	C509	Cancer of the Breast	15q11.2del	
FEMALE	C509	Cancer of the Breast	15q11.2del	
FEMALE	C509	Cancer of the Breast	15q11.2del	
FEMALE	C509	Cancer of the Breast	15q11.2del	
FEMALE	C509	Cancer of the Breast	15q11.2del	
FEMALE	C541	Cancer of the Endometrium	15q11.2del	
MALE	C61	Cancer of the Prostate	15q11.2del	
MALE	C61	Cancer of the Prostate	15q11.2del	
MALE	C64	Cancer of the Kidney	15q11.2del	
MALE	C64	Cancer of the Kidney	15q11.2del	
FEMALE	C679	Cancer of the Bladder	15q11.2del	
MALE	C719	Cancer of the Brain	15q11.2del	
MALE	C719	Cancer of the Brain	15q11.2del	
FEMALE	C80	Cancer, site not known	15q11.2del	
FEMALE	C800	Cancer, site not known	15q11.2del	
MALE	C920	Acute myeloid leukaemia	15q11.2del	
MALE	C920	Acute myeloid leukaemia	15q11.2del	
MALE	D430	Cancer of the Brain	15q11.2del	
MALE	E115	Diabetes	15q11.2del	
FEMALE	E149	Diabetes	15q11.2del	
MALE	F03	Dementia	15q11.2del	
FEMALE	G309	Alzheimer's disease	15q11.2del	
MALE	G35	Multiple sclerosis	15q11.2del	
MALE	I219	Myocardial Infarction	15q11.2del	15q13.3dup_CHRNA7
MALE	I249	Ischaemic heart disease	15q11.2del	
MALE	I259	Ischaemic heart disease	15q11.2del	
FEMALE	I38	Endocarditis	15q11.2del	
MALE	I517	Cardiomegaly	15q11.2del	
MALE	I517	Cardiomegaly	15q11.2del	
MALE	I713	Aortic aneurysm	15q11.2del	
FEMALE	J13	Pneumonia	15q11.2del	
MALE	J181	Pneumonia	15q11.2del	15q13.3dup_CHRNA7
FEMALE	K297	Gastritis	15q11.2del	

MALE	R99	Unspecified	15q11.2del	
MALE	C159	Cancer of the Oesophagus	15q11.2dup	16p13.11.dup
MALE	C159	Cancer of the Oesophagus	15q11.2dup	
MALE	C159	Cancer of the Oesophagus	15q11.2dup	
FEMALE	C169	Cancer of the Stomach	15q11.2dup	
MALE	C189	Cancer of the Colon	15q11.2dup	
MALE	C189	Cancer of the Colon	15q11.2dup	
FEMALE	C221	Cancer of the Liver	15q11.2dup	
FEMALE	C258	Cancer of the Pancreas	15q11.2dup	
MALE	C259	Cancer of the Pancreas	15q11.2dup	
MALE	C259	Cancer of the Pancreas	15q11.2dup	
MALE	C259	Cancer of the Pancreas	15q11.2dup	
MALE	C349	Cancer of the lung	15q11.2dup	
MALE	C349	Cancer of the lung	15q11.2dup	
FEMALE	C349	Cancer of the lung	15q11.2dup	
MALE	C349	Cancer of the lung	15q11.2dup	
FEMALE	C349	Cancer of the lung	15q11.2dup	
MALE	C439	Cancer of the skin	15q11.2dup	
FEMALE	C509	Cancer of the Breast	15q11.2dup	
FEMALE	C509	Cancer of the Breast	15q11.2dup	
FEMALE	C509	Cancer of the Breast	15q11.2dup	
FEMALE	C541	Cancer of the Endometrium	15q11.2dup	
FEMALE	C56	Cancer of the Ovary	15q11.2dup	
FEMALE	C56	Cancer of the Ovary	15q11.2dup	
MALE	C61	Cancer of the Prostate	15q11.2dup	
MALE	C61	Cancer of the Prostate	15q11.2dup	
MALE	C64	Cancer of the Kidney	15q11.2dup	
MALE	C800	Cancer, site not known	15q11.2dup	
FEMALE	C819	Hodgkin's disease	15q11.2dup	
FEMALE	C830	non-Hodgkin's lymphoma	15q11.2dup	
MALE	C859	Non-Hodgkin's lymphoma	15q11.2dup	
MALE	C97	Cancer	15q11.2dup	
MALE	D619	Aplastic anaemia	15q11.2dup	
FEMALE	D860	Sarcoidosis	15q11.2dup	
MALE	G122	Motor neuron disease	15q11.2dup	
MALE	G20	Parkinson's disease	15q11.2dup	
MALE	I219	Myocardial Infarction	15q11.2dup	
MALE	I219	Myocardial Infarction	15q11.2dup	
MALE	I251	Ischaemic heart disease	15q11.2dup	
MALE	I251	Ischaemic heart disease	15q11.2dup	
MALE	I259	Ischaemic heart disease	15q11.2dup	
MALE	I259	Ischaemic heart disease	15q11.2dup	
MALE	I64	Stroke	15q11.2dup	
MALE	I713	Aortic aneurysm	15q11.2dup	
MALE	J189	Pneumonia	15q11.2dup	
MALE	J440	COPD	15q11.2dup	
MALE	J449	COPD	15q11.2dup	
MALE	J449	COPD	15q11.2dup	
MALE	J47	Bronchiectasis	15q11.2dup	
FEMALE	J841	Other interstitial pulmonary diseases	15q11.2dup	
MALE	J841	Other interstitial pulmonary diseases	15q11.2dup	
FEMALE	K550	Vascular disorders of intestine	15q11.2dup	
MALE	K650	Peritonitis	15q11.2dup	
MALE	K700	Alcoholic liver disease	15q11.2dup	
MALE	K709	Alcoholic liver disease	15q11.2dup	
MALE	K746	Cirrhosis of liver	15q11.2dup	
FEMALE	K746	Cirrhosis of liver	15q11.2dup	
FEMALE	X450	Alcohol poisoning, accidental	15q11.2dup	
MALE	X700	Suicide	15q11q13dup_BP3_BP5	
FEMALE	I609	Intracerebral haemorrhage	15q13.3del	
FEMALE	J47	Bronchiectasis	15q13.3del	
MALE	R99	Unspecified	15q13.3del	
MALE	C159	Cancer of the Oesophagus	15q13.3dup	
MALE	C159	Cancer of the Oesophagus	15q13.3dup	
FEMALE	C170	Cancer of the Small Intestine	15q13.3dup	
FEMALE	C189	Cancer of the Colon	15q13.3dup	
FEMALE	C19	Cancer of the rectosigmoid junction	15q13.3dup	
FEMALE	C221	Cancer of the Liver	15q13.3dup	
MALE	C259	Cancer of the Pancreas	15q13.3dup	
FEMALE	C259	Cancer of the Pancreas	15q13.3dup	
MALE	C349	Cancer of the lung	15q13.3dup	
MALE	C800	Cancer, site not known	15q13.3dup	
MALE	I110	Hypertensive heart disease	15q13.3dup	
MALE	I219	Myocardial Infarction	15q13.3dup	

FEMALE	I251	Ischaemic heart disease	15q13.3dup	
MALE	I259	Ischaemic heart disease	15q13.3dup	
MALE	I500	Heart failure	15q13.3dup	
FEMALE	I619	Intracerebral haemorrhage	15q13.3dup	
MALE	X780	Suicide	15q13.3dup	
MALE	B24	HIV	15q13.3dup_CHRNA7	
FEMALE	C159	Cancer of the Oesophagus	15q13.3dup_CHRNA7	
MALE	C159	Cancer of the Oesophagus	15q13.3dup_CHRNA7	
FEMALE	C180	Cancer of the Colon	15q13.3dup_CHRNA7	
FEMALE	C189	Cancer of the Colon	15q13.3dup_CHRNA7	
FEMALE	C19	Cancer of the rectosigmoid junction	15q13.3dup_CHRNA7	
MALE	C20	Cancer of the Rectum	15q13.3dup_CHRNA7	
MALE	C221	Cancer of the Liver	15q13.3dup_CHRNA7	
FEMALE	C259	Cancer of the Pancreas	15q13.3dup_CHRNA7	
MALE	C259	Cancer of the Pancreas	15q13.3dup_CHRNA7	
FEMALE	C259	Cancer of the Pancreas	15q13.3dup_CHRNA7	
FEMALE	C259	Cancer of the Pancreas	15q13.3dup_CHRNA7	
MALE	C259	Cancer of the Pancreas	15q13.3dup_CHRNA7	
MALE	C259	Cancer of the Pancreas	15q13.3dup_CHRNA7	
MALE	C349	Cancer of the lung	15q13.3dup_CHRNA7	
MALE	C349	Cancer of the lung	15q13.3dup_CHRNA7	
FEMALE	C349	Cancer of the lung	15q13.3dup_CHRNA7	
FEMALE	C349	Cancer of the lung	15q13.3dup_CHRNA7	
MALE	C349	Cancer of the lung	15q13.3dup_CHRNA7	
MALE	C439	Cancer of the skin	15q13.3dup_CHRNA7	
MALE	C459	Mesothelioma	15q13.3dup_CHRNA7	
FEMALE	C509	Cancer of the Breast	15q13.3dup_CHRNA7	
FEMALE	C509	Cancer of the Breast	15q13.3dup_CHRNA7	
FEMALE	C509	Cancer of the Breast	15q13.3dup_CHRNA7	
FEMALE	C541	Cancer of the Endometrium	15q13.3dup_CHRNA7	
FEMALE	C56	Cancer of the Ovary	15q13.3dup_CHRNA7	
FEMALE	C56	Cancer of the Ovary	15q13.3dup_CHRNA7	
FEMALE	C579	Cancer of the female genitals	15q13.3dup_CHRNA7	
MALE	C61	Cancer of the Prostate	15q13.3dup_CHRNA7	
MALE	C61	Cancer of the Prostate	15q13.3dup_CHRNA7	
MALE	C61	Cancer of the Prostate	15q13.3dup_CHRNA7	
MALE	C61	Cancer of the Prostate	15q13.3dup_CHRNA7	
MALE	C61	Cancer of the Prostate	15q13.3dup_CHRNA7	
FEMALE	C64	Cancer of the Kidney	15q13.3dup_CHRNA7	
MALE	C64	Cancer of the Kidney	15q13.3dup_CHRNA7	
FEMALE	C64	Cancer of the Kidney	15q13.3dup_CHRNA7	
FEMALE	C64	Cancer of the Kidney	15q13.3dup_CHRNA7	
MALE	C64	Cancer of the Kidney	15q13.3dup_CHRNA7	
MALE	C719	Cancer of the Brain	15q13.3dup_CHRNA7	
MALE	C831	non-Hodgkin's lymphoma	15q13.3dup_CHRNA7	
FEMALE	C844	Lymphoma	15q13.3dup_CHRNA7	
MALE	C845	Lymphoma	15q13.3dup_CHRNA7	
MALE	C845	Lymphoma	15q13.3dup_CHRNA7	
FEMALE	C900	Multiple myeloma	15q13.3dup_CHRNA7	
FEMALE	D758	Other specified diseases of blood	15q13.3dup_CHRNA7	
MALE	G122	Motor neuron disease	15q13.3dup_CHRNA7	
FEMALE	G35	Multiple sclerosis	15q13.3dup_CHRNA7	
MALE	I219	Myocardial Infarction	15q13.3dup_CHRNA7	
MALE	I219	Myocardial Infarction	15q13.3dup_CHRNA7	
MALE	I219	Myocardial Infarction	15q13.3dup_CHRNA7	
MALE	I219	Myocardial Infarction	15q13.3dup_CHRNA7	
MALE	I251	Ischaemic heart disease	15q13.3dup_CHRNA7	
MALE	I255	Ischaemic cardiomyopathy	15q13.3dup_CHRNA7	
MALE	I259	Ischaemic heart disease	15q13.3dup_CHRNA7	
MALE	I259	Ischaemic heart disease	15q13.3dup_CHRNA7	
MALE	I509	Heart failure	15q13.3dup_CHRNA7	
MALE	I607	Intracerebral haemorrhage	15q13.3dup_CHRNA7	
MALE	I64	Stroke	15q13.3dup_CHRNA7	
MALE	I64	Stroke	15q13.3dup_CHRNA7	
FEMALE	I828	Other venous embolism and thrombosis	15q13.3dup_CHRNA7	
MALE	J180	Pneumonia	15q13.3dup_CHRNA7	
FEMALE	J189	Pneumonia	15q13.3dup_CHRNA7	
MALE	J449	COPD	15q13.3dup_CHRNA7	
FEMALE	J841	Other interstitial pulmonary diseases	15q13.3dup_CHRNA7	
MALE	J841	Other interstitial pulmonary diseases	15q13.3dup_CHRNA7	
MALE	J984	Other respiratory disorders	15q13.3dup_CHRNA7	
FEMALE	K551	Vascular disorders of intestine	15q13.3dup_CHRNA7	
MALE	K704	Alcoholic liver disease	15q13.3dup_CHRNA7	
MALE	K709	Alcoholic liver disease	15q13.3dup_CHRNA7	
FEMALE	K859	Other diseases of pancreas	15q13.3dup_CHRNA7	

MALE	K859	Other diseases of pancreas	15q13.3dup_CHRNA7	
FEMALE	N319	Neuromuscular dysfunction of bladder	15q13.3dup_CHRNA7	
MALE	V184	Pedal cyclist accident	15q13.3dup_CHRNA7	
MALE	W190	Unspecified fall	15q13.3dup_CHRNA7	
MALE	X450	Alcohol poisoning, accidental	15q13.3dup_CHRNA7	
MALE	I517	Cardiomegaly	15q24_dup	
MALE	C169	Cancer of the Stomach	16p11.2del	
FEMALE	C259	Cancer of the Pancreas	16p11.2del	2q13dup_NPHP1
MALE	C349	Cancer of the lung	16p11.2del	
FEMALE	E668	Obesity	16p11.2del	
MALE	G931	Anoxic brain damage	16p11.2del	15q13.3dup_CHRNA7
MALE	I110	Hypertensive heart disease	16p11.2del	
MALE	I219	Myocardial Infarction	16p11.2del	
MALE	I509	Heart failure	16p11.2del	
FEMALE	J181	Pneumonia	16p11.2del	
MALE	K709	Alcoholic liver disease	16p11.2del	
MALE	C719	Cancer of the Brain	16p11.2distal_del	
MALE	I259	Ischaemic heart disease	16p11.2distal_del	
MALE	J189	Pneumonia	16p11.2distal_del	
FEMALE	C349	Cancer of the lung	16p11.2distal_dup	
FEMALE	C349	Cancer of the lung	16p11.2distal_dup	
MALE	C349	Cancer of the lung	16p11.2distal_dup	
FEMALE	C80	Cancer, site not known	16p11.2distal_dup	
MALE	I251	Ischaemic heart disease	16p11.2distal_dup	
MALE	J440	COPD	16p11.2distal_dup	
FEMALE	C349	Cancer of the lung	16p11.2dup	
MALE	C719	Cancer of the Brain	16p11.2dup	
FEMALE	C829	non-Hodgkin's lymphoma	16p11.2dup	
MALE	C20	Cancer of the Rectum	16p12.1del	
MALE	C221	Cancer of the Liver	16p12.1del	
FEMALE	C509	Cancer of the Breast	16p12.1del	
MALE	C719	Cancer of the Brain	16p12.1del	
MALE	C900	Multiple myeloma	16p12.1del	
MALE	F019	Vascular dementia	16p12.1del	
MALE	G318	degenerative diseases of nervous system	16p12.1del	
MALE	I219	Myocardial Infarction	16p12.1del	
FEMALE	I251	Ischaemic heart disease	16p12.1del	
FEMALE	I251	Ischaemic heart disease	16p12.1del	
MALE	I269	Pulmonary embolism	16p12.1del	
MALE	I64	Stroke	16p12.1del	
MALE	J180	Pneumonia	16p12.1del	
MALE	J841	Other interstitial pulmonary diseases	16p12.1del	
MALE	X410	Accidental poisoning	16p12.1del	
MALE	C19	Cancer of the rectosigmoid junction	16p12.1dup	
FEMALE	C259	Cancer of the Pancreas	16p12.1dup	
FEMALE	C56	Cancer of the Ovary	16p12.1dup	
MALE	C61	Cancer of the Prostate	16p12.1dup	
MALE	F102	Alcohol Dependence	16p12.1dup	
FEMALE	I219	Myocardial Infarction	16p12.1dup	
MALE	I219	Myocardial Infarction	16p12.1dup	
MALE	W190	Unspecified fall	16p12.1dup	
MALE	C159	Cancer of the Oesophagus	16p13.11.dup	
MALE	C159	Cancer of the Oesophagus	16p13.11.dup	
MALE	C180	Cancer of the Colon	16p13.11.dup	
MALE	C189	Cancer of the Colon	16p13.11.dup	
MALE	C259	Cancer of the Pancreas	16p13.11.dup	
MALE	C329	Cancer of the Larynx	16p13.11.dup	
MALE	C349	Cancer of the lung	16p13.11.dup	
FEMALE	C349	Cancer of the lung	16p13.11.dup	
FEMALE	C509	Cancer of the Breast	16p13.11.dup	
FEMALE	C541	Cancer of the Endometrium	16p13.11.dup	
FEMALE	C56	Cancer of the Ovary	16p13.11.dup	
FEMALE	C56	Cancer of the Ovary	16p13.11.dup	
MALE	C61	Cancer of the Prostate	16p13.11.dup	
MALE	C61	Cancer of the Prostate	16p13.11.dup	
MALE	C679	Cancer of the Bladder	16p13.11.dup	
FEMALE	C710	Cancer of the Brain	16p13.11.dup	
MALE	C719	Cancer of the Brain	16p13.11.dup	
FEMALE	C719	Cancer of the Brain	16p13.11.dup	
MALE	C859	Non-Hodgkin's lymphoma	16p13.11.dup	
FEMALE	D432	Cancer of the Brain	16p13.11.dup	
FEMALE	E785	Hyperlipidaemia	16p13.11.dup	
MALE	G20	Parkinson's disease	16p13.11.dup	
FEMALE	G318	degenerative diseases of nervous system	16p13.11.dup	
MALE	I119	Hypertensive heart disease	16p13.11.dup	

MALE	I219	Myocardial Infarction	16p13.11.dup	
MALE	I219	Myocardial Infarction	16p13.11.dup	
MALE	I251	Ischaemic heart disease	16p13.11.dup	
FEMALE	I48	Atrial fibrillation	16p13.11.dup	
FEMALE	I609	Intracerebral haemorrhage	16p13.11.dup	
FEMALE	J189	Pneumonia	16p13.11.dup	
MALE	J22	Pneumonia	16p13.11.dup	
MALE	J449	COPD	16p13.11.dup	
MALE	J449	COPD	16p13.11.dup	
MALE	J690	Pneumonitis due to food and vomit	16p13.11.dup	
MALE	K550	Vascular disorders of intestine	16p13.11.dup	
FEMALE	K631	Perforation of intestine	16p13.11.dup	
FEMALE	Q446	Cystic disease of liver	16p13.11.dup	
MALE	W15	Fall from cliff	16p13.11.dup	
MALE	X640	Suicide	16p13.11.dup	
FEMALE	C189	Cancer of the Colon	16p13.11del	
MALE	C260	Cancer of other digestive organs	16p13.11del	
MALE	C260	Cancer of other digestive organs	16p13.11del	
MALE	K922	Gastro-intestinal haemorrhage	16p13.11del	
MALE	G35	Multiple sclerosis	17p12del_HNPP	
MALE	X718	Suicide	17p12del_HNPP	
MALE	C349	Cancer of the lung	17p12dup_CMT1A	
MALE	J440	COPD	17p12dup_CMT1A	
FEMALE	J449	COPD	17p12dup_CMT1A	
MALE	C911	Chronic lymphocytic leukaemia	17q12del	
MALE	C349	Cancer of the lung	17q12dup	
MALE	C349	Cancer of the lung	17q12dup	
MALE	I219	Myocardial Infarction	17q12dup	
FEMALE	I251	Ischaemic heart disease	17q12dup	
FEMALE	J151	Pneumonia	17q12dup	
MALE	J189	Pneumonia	17q12dup	
FEMALE	X700	Suicide	17q12dup	
MALE	C20	Cancer of the Rectum	1q21.1del	
FEMALE	G122	Motor neuron disease	1q21.1del	
FEMALE	I420	Cardiomyopathy	1q21.1del	
FEMALE	I609	Intracerebral haemorrhage	1q21.1del	
MALE	J441	COPD	1q21.1del	
FEMALE	J441	COPD	1q21.1del	
MALE	K254	Gastric ulcer	1q21.1del	
FEMALE	C187	Cancer of the Colon	1q21.1dup	
FEMALE	C349	Cancer of the lung	1q21.1dup	
MALE	C349	Cancer of the lung	1q21.1dup	
FEMALE	C56	Cancer of the Ovary	1q21.1dup	
MALE	C719	Cancer of the Brain	1q21.1dup	
MALE	C920	Acute myeloid leukaemia	1q21.1dup	
MALE	I259	Ischaemic heart disease	1q21.1dup	
FEMALE	I639	Cerebral infarction	1q21.1dup	
MALE	J841	Other interstitial pulmonary diseases	1q21.1dup	
FEMALE	G409	Epilepsy	22q11.2del	
MALE	I710	Aortic aneurysm	22q11.2distal_del	
MALE	C159	Cancer of the Oesophagus	22q11.2dup	
FEMALE	C259	Cancer of the Pancreas	22q11.2dup	
FEMALE	C509	Cancer of the Breast	22q11.2dup	
FEMALE	C509	Cancer of the Breast	22q11.2dup	
MALE	C61	Cancer of the Prostate	22q11.2dup	
MALE	C859	Non-Hodgkin's lymphoma	22q11.2dup	
MALE	I251	Ischaemic heart disease	22q11.2dup	
FEMALE	I359	Aortic valve disorder	22q11.2dup	
MALE	J690	Pneumonitis due to food and vomit	22q11.2dup	
MALE	J841	Other interstitial pulmonary diseases	22q11.2dup	
FEMALE	K559	Vascular disorders of intestine	22q11.2dup	
MALE	K746	Cirrhosis of liver	22q11.2dup	
MALE	K802	Cholelithiasis	22q11.2dup	
MALE	R99	Unspecified	22q11.2dup	
FEMALE	C56	Cancer of the Ovary	2q11.2dup	
FEMALE	C859	Non-Hodgkin's lymphoma	2q13del	
MALE	C139	Cancer of the Hypopharynx	2q13del_NPHP1	
MALE	C159	Cancer of the Oesophagus	2q13del_NPHP1	
MALE	C159	Cancer of the Oesophagus	2q13del_NPHP1	
MALE	C160	Cancer of the Stomach	2q13del_NPHP1	
MALE	C187	Cancer of the Colon	2q13del_NPHP1	
MALE	C189	Cancer of the Colon	2q13del_NPHP1	
MALE	C189	Cancer of the Colon	2q13del_NPHP1	
FEMALE	C19	Cancer of the rectosigmoid junction	2q13del_NPHP1	
FEMALE	C19	Cancer of the rectosigmoid junction	2q13del_NPHP1	

MALE	C220	Cancer of the Liver	2q13del_NPHP1	
FEMALE	C221	Cancer of the Liver	2q13del_NPHP1	
MALE	C259	Cancer of the Pancreas	2q13del_NPHP1	15q11.2dup
MALE	C259	Cancer of the Pancreas	2q13del_NPHP1	
MALE	C329	Cancer of the Larynx	2q13del_NPHP1	
MALE	C349	Cancer of the lung	2q13del_NPHP1	15q11.2dup
MALE	C349	Cancer of the lung	2q13del_NPHP1	
MALE	C349	Cancer of the lung	2q13del_NPHP1	
MALE	C349	Cancer of the lung	2q13del_NPHP1	
FEMALE	C349	Cancer of the lung	2q13del_NPHP1	
FEMALE	C349	Cancer of the lung	2q13del_NPHP1	
MALE	C349	Cancer of the lung	2q13del_NPHP1	
MALE	C459	Mesothelioma	2q13del_NPHP1	
MALE	C499	Cancer of connective and soft tissue	2q13del_NPHP1	
FEMALE	C509	Cancer of the Breast	2q13del_NPHP1	
FEMALE	C509	Cancer of the Breast	2q13del_NPHP1	
FEMALE	C509	Cancer of the Breast	2q13del_NPHP1	
FEMALE	C56	Cancer of the Ovary	2q13del_NPHP1	
FEMALE	C56	Cancer of the Ovary	2q13del_NPHP1	
MALE	C61	Cancer of the Prostate	2q13del_NPHP1	
MALE	C61	Cancer of the Prostate	2q13del_NPHP1	
FEMALE	C64	Cancer of the Kidney	2q13del_NPHP1	
MALE	C719	Cancer of the Brain	2q13del_NPHP1	
FEMALE	C800	Cancer, site not known	2q13del_NPHP1	
FEMALE	C851	Lymphoma	2q13del_NPHP1	
FEMALE	C900	Multiple myeloma	2q13del_NPHP1	
MALE	C900	Multiple myeloma	2q13del_NPHP1	
FEMALE	D471	Chronic myeloproliferative disease	2q13del_NPHP1	
MALE	E854	Amyloidosis	2q13del_NPHP1	
MALE	F03	Dementia	2q13del_NPHP1	
MALE	G122	Motor neuron disease	2q13del_NPHP1	
FEMALE	G122	Motor neuron disease	2q13del_NPHP1	
MALE	G231	Other degenerative diseases of basal ganglia	2q13del_NPHP1	
FEMALE	I214	Myocardial Infarction	2q13del_NPHP1	
MALE	I219	Myocardial Infarction	2q13del_NPHP1	
MALE	I219	Myocardial Infarction	2q13del_NPHP1	
FEMALE	I251	Ischaemic heart disease	2q13del_NPHP1	
MALE	I259	Ischaemic heart disease	2q13del_NPHP1	
MALE	I259	Ischaemic heart disease	2q13del_NPHP1	
FEMALE	I604	Intracerebral haemorrhage	2q13del_NPHP1	
MALE	I64	Stroke	2q13del_NPHP1	
FEMALE	I64	Stroke	2q13del_NPHP1	
FEMALE	I802	Phlebitis and thrombophlebitis	2q13del_NPHP1	
MALE	J440	COPD	2q13del_NPHP1	
FEMALE	J849	Other interstitial pulmonary diseases	2q13del_NPHP1	
MALE	J849	Other interstitial pulmonary diseases	2q13del_NPHP1	
MALE	K709	Alcoholic liver disease	2q13del_NPHP1	
MALE	M069	Rheumatoid arthritis	2q13del_NPHP1	
FEMALE	W19	Unspecified fall	2q13del_NPHP1	
MALE	X000	Exposure to smoke	2q13del_NPHP1	
FEMALE	X590	Exposure to unspecified factor	2q13del_NPHP1	
MALE	Y30	Falling, jumping or pushed from a high place, unde	2q13del_NPHP1	
MALE	J849	Other interstitial pulmonary diseases	2q13dup	2q13del_NPHP1
MALE	K922	Gastro-intestinal haemorrhage	2q13dup	
MALE	C119	Cancer of the Nasopharynx	2q13dup_NPHP1	
MALE	C159	Cancer of the Oesophagus	2q13dup_NPHP1	
MALE	C180	Cancer of the Colon	2q13dup_NPHP1	
MALE	C189	Cancer of the Colon	2q13dup_NPHP1	
MALE	C19	Cancer of the rectosigmoid junction	2q13dup_NPHP1	
FEMALE	C20	Cancer of the Rectum	2q13dup_NPHP1	
MALE	C220	Cancer of the Liver	2q13dup_NPHP1	
MALE	C259	Cancer of the Pancreas	2q13dup_NPHP1	
MALE	C260	Cancer of other digestive organs	2q13dup_NPHP1	
MALE	C349	Cancer of the lung	2q13dup_NPHP1	
MALE	C349	Cancer of the lung	2q13dup_NPHP1	
MALE	C349	Cancer of the lung	2q13dup_NPHP1	
MALE	C439	Cancer of the skin	2q13dup_NPHP1	
MALE	C61	Cancer of the Prostate	2q13dup_NPHP1	
MALE	C61	Cancer of the Prostate	2q13dup_NPHP1	
FEMALE	C80	Cancer, site not known	2q13dup_NPHP1	
MALE	C800	Cancer, site not known	2q13dup_NPHP1	
MALE	C809	Cancer, site not known	2q13dup_NPHP1	
FEMALE	C920	Acute myeloid leukaemia	2q13dup_NPHP1	
MALE	C920	Acute myeloid leukaemia	2q13dup_NPHP1	
MALE	C97	Cancer	2q13dup_NPHP1	



MALE	F019	Vascular dementia	2q13dup_NPHP1	
MALE	G122	Motor neuron disease	2q13dup_NPHP1	
MALE	G122	Motor neuron disease	2q13dup_NPHP1	
FEMALE	G20	Parkinson's disease	2q13dup_NPHP1	
MALE	G20	Parkinson's disease	2q13dup_NPHP1	
MALE	I119	Hypertensive heart disease	2q13dup_NPHP1	
FEMALE	I219	Myocardial Infarction	2q13dup_NPHP1	
MALE	I251	Ischaemic heart disease	2q13dup_NPHP1	
MALE	I251	Ischaemic heart disease	2q13dup_NPHP1	
FEMALE	I269	Pulmonary embolism	2q13dup_NPHP1	15q13.3dup_CHRNA7
FEMALE	I269	Pulmonary embolism	2q13dup_NPHP1	
MALE	I269	Pulmonary embolism	2q13dup_NPHP1	
MALE	I38	Endocarditis	2q13dup_NPHP1	
MALE	I429	Cardiomyopathy	2q13dup_NPHP1	
FEMALE	I607	Intracerebral haemorrhage	2q13dup_NPHP1	
FEMALE	I679	Other cerebrovascular disease	2q13dup_NPHP1	
MALE	I839	Other venous embolism and thrombosis	2q13dup_NPHP1	
FEMALE	J180	Pneumonia	2q13dup_NPHP1	
MALE	J180	Pneumonia	2q13dup_NPHP1	
FEMALE	J441	COPD	2q13dup_NPHP1	
MALE	J47	Bronchiectasis	2q13dup_NPHP1	
FEMALE	K704	Alcoholic liver disease	2q13dup_NPHP1	
MALE	K709	Alcoholic liver disease	2q13dup_NPHP1	
FEMALE	K709	Alcoholic liver disease	2q13dup_NPHP1	
MALE	V892	Motor accident	2q13dup_NPHP1	
MALE	I259	Ischaemic heart disease	2q21.1del	
MALE	C029	Cancer of the tongue	2q21.1dup	
FEMALE	C259	Cancer of the Pancreas	2q21.1dup	
MALE	I219	Myocardial Infarction	2q21.1dup	
MALE	K859	Other diseases of pancreas	2q21.1dup	
MALE	R99	Unspecified	2q21.1dup	
FEMALE	C509	Cancer of the Breast	7q11.23dup_distal	
FEMALE	I619	Intracerebral haemorrhage	7q11.23dup_distal	
MALE	J189	Pneumonia	7q11.23dup_distal	
FEMALE	C180	Cancer of the Colon	NRXN1del	
FEMALE	C189	Cancer of the Colon	NRXN1del	
MALE	C439	Cancer of the skin	NRXN1del	
FEMALE	I609	Intracerebral haemorrhage	NRXN1del	
MALE	K266	Duodenal ulcer	NRXN1del	
MALE	C509	Cancer of the Breast	Potocki_Lupski	
MALE	I219	Myocardial Infarction	PWS_dup	
FEMALE	C509	Cancer of the Breast	TAR_del	
FEMALE	C56	Cancer of the Ovary	TAR_del	
MALE	J449	COPD	TAR_del	
MALE	C159	Cancer of the Oesophagus	TAR_dup	
MALE	C169	Cancer of the Stomach	TAR_dup	
FEMALE	C169	Cancer of the Stomach	TAR_dup	
FEMALE	C541	Cancer of the Endometrium	TAR_dup	
FEMALE	C56	Cancer of the Ovary	TAR_dup	
FEMALE	C851	Lymphoma	TAR_dup	
MALE	C859	Non-Hodgkin's lymphoma	TAR_dup	
MALE	C920	Acute myeloid leukaemia	TAR_dup	1q21.1dup
MALE	I711	Aortic aneurysm	TAR_dup	
MALE	J449	COPD	TAR_dup	2q13dup_NPHP1
MALE	J449	COPD	TAR_dup	

